



SPRING 2017

# LIFELINE

COOLEY'S ANEMIA FOUNDATION • LEADING THE FIGHT AGAINST THALASSEMIA

## THOUSANDS ACROSS COUNTRY JOIN CARE WALK 2017



This past May, groups of advocates gathered in many different cities across the U.S. to Care Walk in honor and support of all those living with the challenges of thalassemia. Care Walk is the Cooley's Anemia Foundation's largest annual fundraising event and opportunity to bring together the thalassemia community and its supporters around the country. Funds raised support thalassemia medical research, patient support services, and education initiatives to raise awareness of the genetic blood disorder.

The outpouring of community support is a direct result of the dedication of all of our cherished volunteers who are committed to leading the fight against thalassemia. All of the work our volunteers put into planning their Care Walks, soliciting donations, and educating the community about thalassemia has made Care Walk have the most impact of any single event

of the year for the Foundation. We want to thank each of our volunteers, participants, and donors for all of your support.

This year, Care Walk has a new corporate sponsor—United 7-Eleven Franchisees of Long Island and New York (UFOLI). Many 7-Eleven stores all over Long Island have posters and coin boxes in support of Cooley's Anemia Foundation which will remain in the stores year-round. We want to show our gratitude for their support by returning the support, so we encourage all in Long Island to drop by your local 7-Eleven. Let them know you are a friend of the Cooley's Anemia Foundation and thank them for helping our patients. We thank UFOLI and each of our other sponsors for continuing to help make the Care Walk a success. For more information on joining our next Care Walk, email [mary@thalassemia.org](mailto:mary@thalassemia.org)

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### CAF EXECUTIVE COMMITTEE MEMBERS

**Anthony J. Viola**  
President

**Amy Celento**  
Vice President

**Janet Kwiatkowski, MD, MSCE**  
Medical Advisory Board Chair

**Dean Hernan**  
Treasurer

**Janice Cenzoprano**  
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**Cammie Brandofino**

**Mary Ann Cervoni-Laiia**

**Ralph Colasanti**

**Terri DiFilippo**

**Robert Ficarra**

**Maria Hadjidemetriou**

**DISCLAIMER:** The information in this publication is for educational purposes only and is not intended to substitute for informed medical advice. You should not use this information to diagnose or treat a health problem or disease without consulting a qualified health care provider. The Cooley's Anemia Foundation strongly encourages you to consult your health care provider with any questions or concerns you may have regarding your condition.

## NEUFELD DEPARTS MEDICAL ADVISORY BOARD, KWIATKOWSKI ASSUMES CHAIRMANSHIP

The Cooley's Anemia Foundation is announcing a change in the Chairmanship of its Medical Advisory Board (MAB). Ellis Neufeld, MD, PhD, who has been Chair of the MAB since 2009, has stepped down as of March 1. Janet Kwiatkowski, MD, MSCE has assumed the Chairmanship of this important Board.

### DR. ELLIS NEUFELD

Dr. Neufeld has stepped aside due to leaving his position as Associate Chief of the Division of Hematology/Oncology at Dana-Farber/Boston Children's Cancer and Blood Disorders Center. He has taken up a new role as Clinical Director, Physician-in-Chief and Executive Vice President of St. Jude Children's Research Hospital.

***"It's been a joy and an honor to serve as the head of the Medical Advisory Board for the past several years...The dedication and skill of the Foundation's volunteers and professional staff are really extraordinary."***

— Dr. Ellis Neufeld



"It's been a joy and an honor to serve as the head of the Medical Advisory Board for the past several years," Dr. Neufeld says. "The dedication and skill of the Foundation's volunteers and professional staff are really extraordinary. The scientists and physicians who take time to review Fellowship and Clinical Research Grants for the MAB, and to help offer clinical pathways and information for patients, are unfailingly generous with their time

and expertise because they know what a great job that CAF has done in improving lives for patients with thalassemia."

### DR. JANET KWIATKOWSKI

Dr. Kwiatkowski is the Director of the Thalassemia Program at the Children's Hospital of Philadelphia and Associate Professor of Pediatrics at the University of Pennsylvania Perelman School of Medicine. She received her medical degree from Columbia University College of Physicians and Surgeons in New York and subsequently received MS degree in Clinical Epidemiology from the University of Pennsylvania. Dr. Kwiatkowski completed her pe-

diatric residency training at Johns Hopkins Hospital in Baltimore, MD followed by her pediatric Hematology/Oncology fellowship training at the Children's Hospital of Philadelphia.

"I am pleased and honored to have been asked to assume the Chairmanship of the CAF Medical Advisory Committee and look forward to working with this distinguished group of thalassemia experts," says Dr. Kwiatkowski. "The breadth and depth of knowledge that this Committee possesses makes it an invaluable asset for both the Foundation and the larger thalassemia community. I also look forward to working more closely



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— Dr. Janet Kwiatkowski

with the Foundation and to providing assistance as needed to continue the Foundation's history of disseminating information to the thalassemia community that is medically sound and balanced. Through this collaboration, I hope to further contribute to Foundation's mission of improving the health and well-being of individuals living with thalassemia."

"The Cooley's Anemia Foundation thanks Dr. Ellis Neufeld for his willingness to provide leadership and expertise to the MAB as its Chair, and to his dedication to the entire thalassemia community," says Anthony J. Viola, National President of CAF. "We will miss his involvement but congratulate him on this tremendous opportunity."

"The Foundation feels extremely fortunate that Dr. Janet Kwiatkowski has agreed to assume the Chairmanship of the MAB," Mr. Viola continues. "Her knowledge of and experience with thalassemia and its treatment, combined with her devotion to patients and her research experience in areas related to thalassemia, make her an excellent choice. We are confident that the MAB will continue to grow and advance under Dr. Kwiatkowski's guidance."

# CAF AWARDS \$260,000 FUNDING IN MEDICAL RESEARCH FELLOWSHIPS



CAF is pleased to announce that five new Cooley's Anemia Foundation Medical Research Fellowships and three renewal Fellowships have been awarded for the 2017-2018 grant cycle. The total amount of funding for the 8 research Fellowships is \$260,000.

These Fellowship recipients were assessed on the basis of the quality of the scientific content, the academic accomplishments and future promise of the investigator, the quality of the mentor in the case of postdoctoral fellowships, and, of particular importance, the relevance of the project to the understanding and treatment of Cooley's anemia. The CAF Scientific Review Committee reviewed all applications carefully while adhering to the highest standard for scientifically un-biased reviews and made its recommendations for funding to the CAF Board of Directors, who approved those recommendations at its annual Board meeting.

Dr. Ellis Neufeld, former Chair of the CAF Scientific Review Committee and Medical Advisory Board, says, "This year's CAF Fellowships cover the very broad range of scientific and clinical topics of interest to the thalassemia community. These are cutting-edge investigations, extending from genomics and prenatal diagnosis to iron metabolism to red blood cell development to clinical studies, to fulfilling the promise of gene therapy. We are excited to see what the investigators learn in these important experiments."

## FIRST YEAR FELLOWSHIP RECIPIENTS

The following individuals have been awarded new Fellowships for this grant cycle. Fellowships are awarded for one year with renewal for a second year contingent upon review of progress.

**Nikoleta Psatha, PhD**, will conduct research in gene editing through her study, "Two innovative approaches for genome editing in beta-thalassemia." In the first approach she will produce simultaneously two fetal hemoglobin activating events in one genetic vector designed for two genomic targets with a CRISPR/Cas9 Nuclease approach. This should increase the

## CAF THANKS JOE DITRAPANI FOR HIS YEARS OF SERVICE

As some of you may know, the Order Sons of Italy in America (OSIA) is the oldest and largest national organization for men and women of Italian heritage. Established in 1959, the Sons of Italy Foundation (SIF) has raised more than \$163 million over the years for educational scholarships, national charities, medical research, cultural preservation, disaster relief, and other special projects.



Joseph J. DiTrapani comes from a legacy of proud Italian Americans as well as Son of Italy members. Two of these members happen to be his parents, who gave Joseph his first introduction to OSIA. He officially became a member of the Order Sons of Italy in 1975. In 1981, Joseph became State President of OSIA's Grand Lodge of New York and has served two terms as OSIA's National President from 2009-2013. Since 2013, Joseph has served as the President of the Sons of Italy Foundation.

As if that's not impressive enough, Joseph, along with the state and local lodges, has relentlessly devoted time throughout the year to raise money for the SIF's national charities with Cooley's Anemia Foundation being one of the primary recipients. Not only has Joseph raised money for charity, but to date SIF has awarded nearly \$61 million in scholarships to students of Italian American descent.

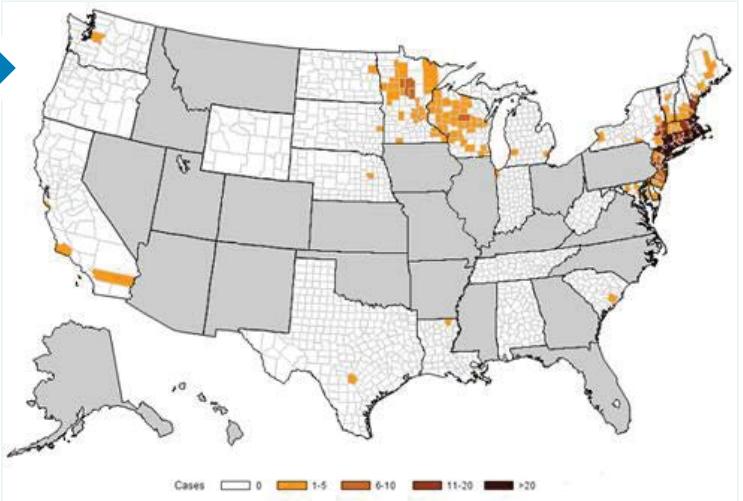
Cooley's Anemia Foundation is very near and dear to SIF due to the fact that this disorder frequently affects Italian Americans. Joseph and the Sons of Italy Foundation became familiar with CAF due to our mutual location in New York City, and CAF was adopted as an official SIF national charity in 1987. "We support CAF because thalassemia is a condition that afflicts thousands of people of Mediterranean descent, notably Italian Americans. We want to do our part to help our brothers and sisters who are suffering from this disease and ultimately find a cure," says Joseph.

In order to raise funds for CAF, OSIA lodges organize fundraising events that occur on a local and state level. The state lodges present their fundraising at the Biennial National Convention and all of the proceeds are collectively sent to CAF directly from SIF. To date, OSIA has contributed close to \$2 million to the Cooley's Anemia Foundation. This prominent organization's main hope for CAF's patients and supporters is to raise enough money to find a cure for this challenging disorder.

## INFECTION AND THALASSEMIA

Although infection-related fatalities in thalassemia are not common, they can occur. We asked Ellis Neufeld, recent Chair of CAF's Medical Advisory Board, to provide some reminders based in medical evidence that people with thalassemia should keep in mind. These include:

- 1.** Persons with no spleens, including thalassemia patients who have had splenectomy, are at increased risk of a certain group of dangerous bacterial germs including pneumococcus, meningococcus, and hemophilus influenza (which is unrelated to flu virus, but same word).
  - a. Fortunately, there are vaccines for these germs. Every splenectomized thalassemia patient at all ages should have up to date vaccines including PCV-13 (Prevnar 13), PPV 23 (Pneumovax), HIB vaccine (hemophilus), and Menactra. The rules about "up to date" are changed by CDC every few years. If you haven't had updates in a few years, please check with your physician immediately to catch up.
  - b. Unfortunately, some forms of pneumococcus are NOT covered by the vaccines.
  - c. Therefore, every fever over 101.5F (38.5C) in splenectomized patients should be considered an emergency. Different physicians have slightly different approaches for this problem, but no high fever can be safely ignored with no spleen.
    - i. Even if other household contacts have a nonbacterial illness (like influenza virus), the splenectomized patient should be seen emergently for high fever. This is true even if the circumstances are inconvenient (e.g., Thanksgiving weekend, traveling on vacation, etc.)
    - ii. In general, this emergency visit should include a blood culture, CBC/diff, and a dose of strong antibiotics awaiting the blood culture results.
    - iii. If a patient seems or feels particularly ill and had no spleen, he or she should also be seen urgently, even without a very high fever. But if the fever is very high, he or she should be seen even if s/he feels otherwise ok.
- 2.** The spleen also is the primary filter for a class of protozoal germs that includes malaria (all forms) and babesiosis. In the US, babesiosis is now famous for being possible to acquire from transfusions, but it is endemic to the coastal Northeast US from Cape May through Massachusetts, and to areas around Maine, New Hampshire, the lakes of Wisconsin and Minnesota, among other places. It is carried by the same deer ticks that carry Lyme disease. Precautions at the beaches (such as ,don't go in the dune grass with shorts on, use insect repellent wisely, etc.) can reduce the risk.



Map showing cases of babesiosis in US.

**3.** Some germs (not the big three noted above for the spleen) grow best when they can get iron from their environment. Paradoxically, chelators, especially deferoxamine (Desferal), help get iron INTO these germs, and this is why we strongly suggest being off chelator at least at the beginning of illness with fever until evaluation is completed, and these germs are ruled out. Particularly bad actors among these germs can include Listeria, Vibrio, and Klebsiella. For these kinds of germs, warnings that come about food borne infections make sense to heed. Probably everyone should get pasteurized milk products, but raw milk particularly carries Listeria, and therefore raw milk (at farms or in some cheeses) should be avoided. Raw seafood, but not cooked seafood, can carry Vibrio. Raw beef (such as steak Tartare) can carry toxoplasmosis. If a person on chelators has unexplained fevers or feels particularly ill, it is very important to tell your physician if you had any of these exposures, and in general any travel.

If you have specific questions about areas not addressed here, please ask your physician. We welcome comments and queries to CAF as well (c.butler@thalassemia.org).

### IN MEMORIAM BENEDICT A. JACOBELLIS 1947-2016

CAF is saddened by the passing of Benedict "Benny" Jacobellis, a dedicated member of the CAF Board of Directors, the Executive Committee, and Vice President of the CAF Suffolk Chapter. Ben was a wonderful man who had a tremendous commitment to the thalassemia community. We are so grateful for his years of service and he will be sorely missed by all who knew him.



# CAF WELCOMES NEW DEVELOPMENT DIRECTOR, HENRY ACKERMANN

Hello, family and friends of the Cooley's Anemia Foundation! I am so happy to be joining you as we work together to advance the treatment and cure for this fatal blood disorder, enhance the quality of life of patients, and educate the medical profession, trait carriers, and the public about thalassemia. Your generosity makes this possible. Thank you for all you do!

Beginning with this issue, we will introduce you to, or remind you of, the various ways you can contribute to our cause. We, of course, gladly welcome traditional cash donations, but there are many other choices available to us. All of these other opportunities fall under the broad heading of "Planned Giving."

We often think of "planned giving" as naming charities in our wills and bequests. This is a form of "planned giving," but there is so much more! Gifts of securities, real estate, art, jewelry, life insurance, trusts, annuities, and other forms of "gift planning" that require more than simply writing a check, are others. Many of these gifts require the assistance of a qualified financial advisor or an attorney, but others can be part of your charitable giving plan by simply designating Cooley's Anemia Foundation as the recipient. In all cases, we are here to guide you.

## AMAZON SMILE

This may be the easiest way to give to the Cooley's Anemia Foundation without writing a check to us! If you buy anything online through Amazon, this is for you! Just go to [smile.amazon.com](https://smile.amazon.com) and follow the instructions. You will either set up an account and designate Cooley's Anemia Foundation as your charity or, if you already have an Amazon account, you will simply designate Cooley's Anemia Foundation as your charity of choice. It is that simple! After that, just order what you want and the AmazonSmile Foundation will donate 0.5% (half a percent) of the purchase price from your eligible purchases.



**Start with a Smile at  
[smile.amazon.com](https://smile.amazon.com) this Holiday Season**

Support us and find all of your holiday essentials including gifts, decorations, and more.

You shop. Amazon gives.

**amazon smile**

From that point on, when you want to order something through Amazon, just go through <https://smile.amazon.com> and 0.5% of your purchases will be sent to Cooley's Anemia Foundation automatically. Remember that the donation is actually coming



from Amazon, not you, so you get no tax benefit – just the satisfaction of knowing you helped us out!

And there is another way you can help. While 0.5% does not seem like much (50 cents for every \$100 spent), it can add up. Imagine if your family and friends joined you in this effort? Imagine if your employer, church, business, or civic club made their purchases through Amazon Smile – we might be talking significant numbers. So spread the word!

## IRA CHARITABLE ROLLOVER

This type of planned gift is not for everyone, but for those who qualify, it can be a terrific way to make a significant contribution!

It's called an Individual Retirement Account Charitable Rollover and here's how it works: Anyone who has set up an IRA is required to start withdrawing a percentage of the account when he/she reaches age 70 1/2 (and do so annually every year thereafter). How much is required to be withdrawn is dependent upon age and total amount in the account. The Internal Revenue Service provides an actuarial chart to determine the amount. With the exception of a Roth IRA, the amount that is withdrawn is taxable income. But suppose there was a way to withdraw the required amount (and even more) without paying any income tax on it and help the Cooley's Anemia Foundation at the same time?

The answer is taking advantage of the IRA Charitable Rollover! As of December 2015, this provision became a permanent part of the tax code, so, anyone 70 1/2 or older can rollover (contribute) the minimum required withdrawal or a larger amount (up to \$100,000 per year!) directly to the Cooley's Anemia Foundation. By having the amount donated directly from your IRA account by the account manager and sent by her/him to the Cooley's Anemia Foundation, you avoid paying any income tax on the amount withdrawn. Win/win! You do not pay tax on the withdrawn amount and we have the benefit of your gift!

Please remember that only the IRA account manager can perform this transaction. If you accept the withdrawal, you must pay income tax on it, even if you subsequently donate it to charity. Using the IRA Charitable Rollover does not permit you to take a charitable deduction on the amount withdrawn (that would be double dipping).

If you consider taking advantage of the IRA Charitable Rollover, please consult your financial advisor to determine if this is what is best for your particular situation.

And there you have it. In subsequent newsletters we will explore other options for you to consider when determining how you can plan your giving to further the good work of the Cooley's Anemia Foundation. Thanks, again, for all you do.

## CAMPS FOR CHILDREN WITH THALASSEMIA

**CAF is happy to provide information about camps for children with serious medical conditions including thalassemia. This is provided for informational purposes so that parents/patients can know about possible options they may wish to consider. CAF does not endorse one camp over another.**

### THE PAINTED TURTLE

The Painted Turtle is a camp in Lake Hughes, California for children with special medical conditions and their families. They host a variety of camps throughout the year and will be hosting a summer camp for children with thalassemia, hemophilia and Von Willebrand disease from July 22-27.

The Painted Turtle has a full-time medical staff and a 24-hour medical center to ensure that children receive all necessary treatments for the duration of the camp. Any child who meets the camp's medical requirements and receives physician approval will have the opportunity to attend. Every camper attends free of charge.

To apply, visit [thepaintedturtle.org](http://thepaintedturtle.org) or contact the Camper Admission Office at 661-724-1550.

### THE DOUBLE H RANCH

The Double H Ranch is a camp located in Lake Luzerne, NY (in the Adirondack Mountains) for children with special medical conditions including thalassemia.

The Ranch offers seven 6-day residential sessions between June and August. There is an on-site medical team and a 24-hour medical center to ensure that children receive necessary medical treatment. Any child who meets the camp's medical requirements and receives physician approval will have the opportunity to attend. Every camper attends free of charge.

The Double H Ranch also offers a series of Family Programs in addition to its Summer Camps. These programs may be ideal for children who would like to experience the camp environment, but who are not yet ready to leave their families behind. The Family Programs are designed for campers with serious medical conditions, ages 5-21 years old, along with accompanying family members. Double H Ranch is planning a Hematology/Oncology Family Weekend this May 5th-7th. Campers and their families are invited to attend this session free-of-charge. Admission to the program is first-come, first-served, and the deadline for applying to this program is April 21st, 2017.

Further information about these programs is available on the Double H Ranch website at: [doublehranch.org](http://doublehranch.org).



### CAMP KOREY

Camp Korey is a camp located in Mt. Vernon, Washington for children living with life-altering medical conditions. They offer a series of summer camp sessions for patients with general diagnoses including thalassemia. Campers between the ages 7-16 can come and enjoy weeklong sessions of fun and adventure. Camp Korey is free of charge to all campers. Further information about this program is available on the Camp Korey website at: [campkorey.org](http://campkorey.org).

### HOLE IN THE WALL GANG CAMP

The Hole in the Wall Gang is a camp located in Ashford, CT for children with special medical conditions. They offer a series of summer camp sessions for patients with General Diagnoses – including Thalassemia – throughout the summer. Campers must be between the ages of 7 and 15 to participate. The Hole in the Wall Gang has a full-time medical staff and a 24-hour medical center to ensure that children receive any necessary treatments while they are at camp. Any child who meets the camp's medical requirements and receives physician approval will have the opportunity to attend. Every camper attends free of charge. Further information about this program is available on the Hole in the Wall Gang website at: [holeinthewallgang.org](http://holeinthewallgang.org).

### ROUNDUP RIVER RANCH

Roundup River Ranch is located along the Colorado River in the heart of the Rocky Mountains. Roundup River Ranch serves children ages 17 and under who have been diagnosed with a serious illness including thalassemia. This camp provides summer sessions based on each camper's medical needs as well as several family camps. The medical team is filled with nurses and physicians who specialize in caring for children who have the medical conditions they serve. Nurses and doctors are available and on call 24 hours a day while campers are on site. For further information about this program, please visit: [roundupriverranch.org](http://roundupriverranch.org).

### VICTORY JUNCTION

Victory Junction is a camp located in Randleman, North Carolina. Victory Junction caters to children with chronic medical conditions or serious illness including thalassemia. This camp is available to children ages 6-16. Victory Junction provides summer sessions, family weekends, and young adult weekends, which focus on individuals from 18-22 years of age who have aged out of traditional camp programs. Victory Junction is free of charge to all children and families. Victory Junction's medical team carefully reviews each child's medical record to ensure that all activities the camper participates in are medically appropriate. For further information about this program, please visit: [victoryjunction.org](http://victoryjunction.org).

## NORTH STAR REACH

North Star Reach is a camp located in Pinckney, Michigan. North Star Reach serves children with serious health challenges including thalassemia. This camp offers summer sessions for children ages 7-15 from June to August. This camp also provides Teen Week for campers that are 16 and 17 years old. Children and their families are able to enjoy this experience free of charge. For further information about this program, please visit: [northstarreach.org](http://northstarreach.org).

## CAMP HOLIDAY TRAILS

Camp Holiday Trails is located in Charlottesville, VA and welcomes children ages 7-17 with special medical conditions including thalassemia. The 75-acre campground includes a medical facility and pharmacy, as well as medical staff of over 50 physicians, nurses, pharmacists, medical students, residents and volunteers. Camp Holiday Trails also offers sessions for families, allowing family bonding time through fun activities. For more information and to apply, visit [campholidaytrails.org](http://campholidaytrails.org).

## CAMP BOGGY CREEK

Camp Boggy Creek is a camp located in Eustis, Florida. This camp provides a free and medically sound camp environment for children living with serious illnesses including thalassemia. Children ages 7-16 with certain specified medical conditions are eligible to apply. Camp Boggy Creek offers weekly summer sessions for children and family retreat programs in the spring and fall. Camp Boggy Creek provide full time physicians and nurses while campers are on the property that specializes caring for children with serious medical conditions. For more information, please visit: [boggycreek.org](http://boggycreek.org).

## FLYING HORSE FARMS

Flying Horse Farms is located just outside of Mt. Gilead, Ohio. The camp facilities are spread over 200 wooded acres and include 22 camp buildings two lakes. In addition to a 24-hour medical facility with a full medical staff, the camp features a boating/fishing harbor, vegetable garden, amphitheater, and archery range among other activities. Children from age 8-15 with a variety of medical conditions, including thalassemia, are welcome to apply. Campers attend free of charge. For more information, visit: [flyinghorsefarms.org](http://flyinghorsefarms.org).

## THE MAN BEHIND THE NAME

**Thomas B. Cooley, M.D.**

(June 23, 1871—October 13, 1945)



People often ask why the major form of thalassemia is known as Cooley's anemia. Thalassemia major is widely referred to as Cooley's anemia in reference to Dr. Thomas Benton Cooley, the renowned researcher who discovered the disorder.

Thomas Benton Cooley was an American physician specializing in pediatrics and hematology. He worked tirelessly to treat illnesses causing the high child and infant mortality rates found in the U.S. and globally in the early 20th century.

After completing medical school at the University of Michigan in 1895, he went on to complete specialized training in hygiene and contagious diseases at Boston City Hospital and in German clinics. Cooley then led the effort to treat individuals infected with rabies as the lead physician of the Pasteur Institute at the University of Michigan from 1903-1905.

Cooley made one of his biggest contributions to the dramatic reduction of infant deaths as the medical director of the Babies' Milk Fund in Detroit. His increasing interest in and dedication to the improved health of children grew, leading him to serve as Assistant Chief of the Children's Bureau of the American Red Cross in France during World War II. During his time in France, he led several projects including: the implementation of a school of public health for children, the establishment of a pediatric hospital, the development of a boarding school for children orphaned by war, and the foundation of a training school for visiting housekeepers who were tasked with visiting impoverished children and ensuring proper hygiene and diet. Cooley's list of accomplishments in France is truly remarkable, inspiring the government of France to award him with the Legion of Honour in 1924 to recognize his impact in the lives of French children.

After returning from France in 1921, Cooley served as the head of pediatrics at the Children's Hospital of Michigan for the following two decades. He soon began an investigation on a form of childhood anemia, noting similarities in bone change among four children of Italian and Greek heritage. Cooley presented his findings to the American Pediatric Society in 1925, naming this disorder erythroblastic anemia, now known as Cooley's anemia. His research is considered one of the most significant contributions to hematology and laid the groundwork for thalassemia research and treatment options in the following decades.

Cooley continued his dedication to children's health and wellbeing by co-founding the American Academy of Pediatrics in 1930, and by serving professor of pediatrics at Wayne State University College of Medicine for nearly a decade beginning in 1936. Cooley died in October 13, 1945.

The Cooley's Anemia Foundation proudly bears the name of Dr. Thomas Benton Cooley in honor of his preeminent contributions to the livelihoods of individuals with thalassemia, and as a source of continued inspiration to the new crop of dedicated medical researchers leading the quest for a cure.

## In Memoriam

We regretfully report the loss of the following Cooley's Anemia Patients and extend our sympathies to their friends and family.

- David Brady
- Kristina Migliore
- Jeffry DiBlasi
- Michael Moussa
- Mark Lepore
- Steven Srisavat
- Hrisafina ("Nina") Mariettos
- Eleni Valusek

## PATIENT PROFILES

### MEET BRIANNA: HIGH SCHOOL SENIOR AND CARE WALK CAPTAIN



I am #ThrivingWithThal  
-Brianna, Senior in High School

**Brianna is an inspiring young leader in the thalassemia community, and CAF thanks her for serving as a Care Walk team captain and for sharing her story.**

in high school, I will be graduating this June and going to college in the fall. I have a sister and a brother, and my household consists of my mom, grandmother, me, and my sister. We are all originally from New York City, but we moved to Bushkill, Pennsylvania in 2007.

I was originally diagnosed with beta thalassemia minor, but it recently advanced to thalassemia intermedia. I also have the alpha thalassemia trait, which I inherited from my father. However, I have hereditary persistence of fetal hemoglobin, so the effects of the thalassemia is less significant than it would normally be.

I found the Cooley's Anemia Foundation in 2016 when I was having abdominal pains and came to find out they were gallbladder complications from the thalassemia. After that incident, I became much more interested in thalassemia and how it affects different people.

This May, my mother and I decided to host our very own Care Walk as Team Bushkill Rocks. This was my first time Care Walking for thalassemia, and it was very exciting! Although I was very busy this spring with senior events and AP testing, I was committed to making Care Walk a success. Our Care Walk took place at my high school, East Stroudsberg High School North, on May 7. I reached my goal of raising \$1,000 for CAF!

I think it is important to raise thalassemia awareness because not many people have heard of the disease and how it affects people. More awareness could possibly result in a cure for thalassemia in the future. Many people do not support disorders they haven't heard about, instead they opt for well-known illnesses; however, I think all disorders, rare included, deserve recognition and support. I Care Walk to offer my support for other people affected by thalassemia around the world in hopes of a cure one day.

For anybody affected by thalassemia, I think it is always important to have a support system, such as your family, to work together to help minimize the symptoms. You should regularly

visit your hematologist or doctor who specializes in blood disorders to monitor the thalassemia and your hemoglobin. For example, when I had my gallbladder crisis back in 2016, having my mother and grandmother there was very comforting and had I ignored the pain, I would have never found out that I had advanced to intermedia. Maintaining your health or child's health is also critical, like exercising and eating fruits and vegetables. Your lifestyle habits should have a positive impact on the thalassemia.

### DANIELLA MACOLINO: ACTRESS LIVING HER DREAMS



I am #ThrivingWithThal  
-Daniella, Actress

**Daniella Macolino is a 24 year old actress who pursues her dreams each day while living, and thriving, with thalassemia major. CAF thanks Daniella for allowing us to share her inspiring story.**

When I was four months old, my family and I were on vacation in Vermont when my mom noticed I was turning yellow after my older brother took me out in the snow. She overlooked it, thinking I may have just been tired from the traveling. But the next day I was looking worse with bags under my eyes, so my parents rushed me to the hospital. The doctor did some tests, and broke the news to my parents that I have thalassemia major and I would need a blood transfusion right away and would need to continue receiving transfusions every two weeks for the rest of my life.

My mother was born and raised in France, and she came to America not speaking any English. Neither of my parents had any knowledge that they carried the thalassemia trait and had never even heard of it before my diagnosis. Since then, my parents have become my biggest advocates and made sure I grew up with an amazing support system of friends and family. My siblings, Paul, Joseph, and Victoria, understand what I go through and don't look at me any differently.

Living with thalassemia has been difficult, especially as a teenager. I wanted to be like my friends—go out and have sleepovers—but couldn't because I had to be home to take my Desferal injection. This nightly routine requires me to sleep with a needle pumping medication into my body to remove the iron buildup from receiving blood transfusions every two weeks. I hated it, and it was very inconvenient for me. It was frustrating waking up with bruises on my legs and arms from the needle being in all night.

Beyond that, it was hard trying to fit in while feeling so differ-

ent because I had an illness and I had to go to the hospital frequently. I only told a few of my closest friends that I had this disorder. Now that I am older and I am used to it, I am very open about sharing my experience with everyone. This illness isn't going to define who I am, but it will make me stronger and I know that's why people are inspired by my story!

The Cooley's Anemia Foundation has been so amazing to me and my family. I can contact the Foundation anytime I have questions or need anything and they are there for me. The amount of thanks I want to give them, I can't even put into words! I have been able to meet the most amazing people by attending Foundation conferences and other events. We are a true community, supporting each other's passions, goals, and dreams.

It is so important to get involved with and support the Foundation, especially if you or someone you know has thalassemia. I know I can speak on behalf of the patients when I say that getting involved is worth it because we are raising awareness of our disorder as well as funds for medical research. One day, there will be a cure and I believe it will come sooner than expected!

"Choose a job you love, and you will never have to work a day in your life." -Confucius

I believe that no matter what, everyone should follow their dreams and no obstacle should stand in the way of accomplishing that! Those who know me know that I am pursuing an acting career and I have been for a while now. I don't know exactly what it is about being in front of the camera or on stage that makes me feel amazing... but I know that I could not live without acting.

I've had to sacrifice so many things in order to keep doing what I love, but I'm no stranger to challenges! This is what I want to do and I will never give up. All the hard work is paying off because each year, I find myself doing better and better in my acting career. From January to May of this year I played a leading role in an independent feature film called "The Prey"—look out for it in October! And in August, I will be in a short horror film which I am really excited about. Tonight, I appear in an episode of "The Perfect Murder" on the ID channel. It is so crazy to even think that I have made it this far! Even though I put myself down every now and then thinking that I could be doing more, I am still really proud of what I have accomplished so far.

My advice to other thalassemia patients is to not look at yourself any differently than anyone else. Live your life day by day. Do what you love and don't listen to anyone who puts you down, because at the end of the day you are a strong individual capable of doing what you want!

Just always remember to be safe and take care of yourself. It is important to be compliant by doing your chelation EVERY DAY and going to get blood transfusions. If you're tired or feeling ill, don't put it to the side! Your health comes first no matter what.

Thalassemia is rare and I know many people have not heard about it, but it is just as dangerous as any other illness. Every patient is different and there are complications. Please raise awareness and if you have never gotten your blood tested, visit your doctor to see if you carry the trait.

## LISA GUIDICE: PROUD JERSEY GIRL #THRIVINGWITHTHAL

**Lisa Falco Guidice is an active member of the CAF patient community, and we thank her for sharing her inspiring story.**



*I am #ThrivingWithThal  
-Lisa Guidice*

I am a proud Jersey girl, born in Summit and raised in Florham Park. After being diagnosed with thalassemia major at 6 months old, my mother did everything she could to ensure that I lived a normal childhood. My favorite thing to do growing up was to follow my sister around and do whatever she was doing with her friends. Although I had to get blood transfusions every 2 weeks, my mom made me feel like having thalassemia did not define me and tried to downplay the role that this disorder had in my life. At the same time, she was very strict about where I could go and what I could do, so I was definitely sheltered. When I was three years old, I started using the Desferal pump. This is a special device that pumps medication into my body to reduce the amount of iron in my blood. It was challenging to have to sleep with a needle in my stomach every night, but I would say I had a very happy childhood overall.

Unfortunately when I was 15 years old, I had two heart attacks and became a diabetic but I didn't let this discourage me. I pushed through, kept up with my medication and blood transfusions, and led a pretty normal life. Now I am in my 13th year of marriage and I am happier than ever! My family and my friends are what push me to never give up. I am striving to live as long as possible to see my niece and nephew grow up, get married, and become successful.

Cooley's Anemia Foundation has been there for me through it all. The Foundation has provided me with many different resources such as helping out with insurance issues, introducing me to a community of people just like me, and providing me with a huge support system. My experience has taught me that thalassemia is not a death sentence. As long as you are being responsible and taking your required medications, you are capable of living a beautiful and long life. All my life I was told by doctors that I would never make it to adulthood but I am now 40 years old and proud! I wake up every day joyful to be alive and to create new memories.

For those who are going through what I go through, I would like to let you know that this disorder does not have to define you or break you down. Even though thalassemia will always be a part of you, it is not who you are. Live your life the way you see fit and do the things that make you happy. Make sure to always surround yourself with positive energy and with those who care for you. You are not alone in your journey.

## VOLUNTEER SPOTLIGHT

CAF launched our “Volunteer Spotlight” Series last year to highlight the stories of some of the many selfless volunteers who play a major role in making our work possible. We are truly grateful for all of the love and heartfelt effort each of our volunteers puts into organizing awareness events, blood drives and fundraisers, and we thank them for their consistent commitment to improving the lives of individuals with thalassemia.

### CAMMIE BRANDOFINO

Cammie Brandofino has been a dedicated volunteer for CAF since 1978 and became a board member in 1985. She currently serves on the CAF National Executive Committee and the Board of the CAF Staten Island Chapter. Her fellow CAF Executive Committee Member, Terri DiFilippo, says, “She is never too busy or too tired to work tirelessly for all with Cooley’s anemia. Her vision is clear—help the patients in their quest to live the best possible life while enduring the vigorous routine of chelation.” We thank Cammie for her dedication to this vision, and for allowing us to share her story.

**“Cammie is never too busy or too tired to work tirelessly for all with Cooley’s anemia.”**

– Terri DiFilippo, CAF Executive Committee Member



My name is Cammie Brandofino and I became involved with the CAF after my daughter Danielle was diagnosed with Cooley’s anemia in 1978. At the time, there wasn’t a chapter available on Staten Island so I would travel to the Brooklyn Chapter to attend meetings. Once I began attending these meetings, I met a wonderful woman named Terri DiFilippo. Our children didn’t go to the same hospital for transfusions so I only saw her at meetings and functions hosted by the Brooklyn

Chapter at that time. When many of the parents living in Brooklyn moved to Staten Island, it was time for us to open a chapter there. We did so and received our charter in 1985. Terri was still living in Brooklyn and we saw each other at National Executive Committee meetings. She moved to Staten Island several years later and became an integral part of the Staten Island Chapter as she is today.

When Danielle was first diagnosed, I felt lost and terrified and knew that I had to find out more about this disorder from others more experienced in dealing with the day to day problems



Cammie and the Pink Ladies

arising from treatments, hospitalizations, etc. My husband and I and the rest of the family needed to help Danielle cope with what was happening to her. Our lives were changed and so was hers. Volunteering with CAF has kept me focused on what’s important in life. It has helped me deal with the realities of having a child living with Cooley’s and has been a most rewarding experience. On this journey, I have been blessed to work with many other parents facing the same difficulties which has created an immense bond between us. We would reach out to each other, compare notes on what new treatments were on the horizon, and give each other courage when we found out yet another patient had passed away. Thank God those days have given way to healthier patients living a wonderful quality of life into their 50’s and 60’s.

One of my favorite fundraising events would have to be our annual Fashion Show held on Staten Island. The patients would be the stars of the show, they looked forward to it every year and were so proud to strut their stuff. A few years ago we held a “Miracle Babies” Fashion Show where we had the patients and their children walk the runway. There wasn’t a dry eye in the place. One of the most rewarding functions of our chapter and one in which our volunteers enjoy the most is when we give stipends to the patients involved in our chapter to help defray some of their out of pocket medical expenses. When we present them with checks at our Christmas meeting, they are so grateful for whatever we can give them and our members are thrilled that their fundraising efforts have been instrumental in helping them.

Fortunately, family and friends have kept our chapter going for over 30 years but a few years ago it was in danger of folding because of dwindling numbers of volunteers. Suddenly, we had a new group of younger, energetic and selfless women who came together to re-create a vibrant, still viable chapter. They have come up with some creative fundraising ideas and have helped to activate awareness in our community.

Our calendar over the past several months and going forward are filled with wonderful events. We’ve had blood drives, a DooWop Entertainment/Dance luncheon, Cosmic Bowling, Zumba Exercise fundraisers, and Community Outreach. We just held our annual Golf Outing and 2017 Care Walk, and that was only the first 6 months of 2017!

We are very fortunate for our patients living on Staten Island and Brooklyn that come and participate in our events. They are the faces our volunteers want to personally get to know who they’re helping and why. Cooley’s Anemia Foundation will forever be a major part of my life.

## MARIA SARADPON AND ERICA MARTINEZ

**Maria Saradpon and Erica Martinez may be fairly new volunteers, but they have already made a big impact on thalassemia awareness by organizing their annual World Thalassemia Day fundraiser for the second year in a row, and re-launching the CAF California Chapter. CAF thanks Maria and Erica for allowing us to share their story, which can also be found on their advocacy website, [thalpalsandiego.com](http://thalpalsandiego.com).**

Our names are Erica Martinez and Maria Saradpon. We are the parents of two incredible little girls who happen to have thalassemia. Ella Martinez is eight years old and Kamila Saradpon is two.

**"When 'i' is replaced by 'we' even illness becomes wellness."**

– Maria Saradpon quotes Malcolm X



Erica Martinez (left),  
Maria Saradpon (right)

We met at Rady Children's Hospital with the help of our former nurse practitioner, Roberta. I (Maria) was a mess. My husband and I just found out a couple months prior that our 5 month old baby girl had a serious chronic illness and would need blood transfusions every 3 weeks for life. She would have surgery the following week to insert a port in her chest because her last blood transfusion took 10 pokes to finally start. A lot for new parents to accept to say the least.

Anyway, bless this nurse's heart- she knew I needed someone to tell me it would all be alright; that I would find comfort in knowing I wasn't alone. She gave me Erica's phone number and encouraged me to call.

Erica had been asking to be connected with other thal parents for years. It never happened, so she eventually stopped asking. Then one day Roberta told her that there was a mom she wanted her to meet. She explained that this family just started treatment and seemed like they could benefit from having another family to connect with. Erica gladly gave permission to release her contact information, and waited patiently for a call.

I didn't call. I don't know why, I guess I was nervous? "Hi! My daughter has thalassemia too. Can we be friends?" Seemed awkward to me any which way I were to say it. Besides that, we had infant surgery to mentally prepare for and a regular blood transfusion schedule to adjust to. I put it off, days turned into weeks, and weeks turned into months. Roberta moved away. I ended up losing the paper I'd doodled her number down on, and thought that was the end of that.

Four months later at one of Kamila's transfusion days, our case manager Beth came up to me and said that Erica was still very

interested in meeting if I was up for it. I apologized for never calling and admitted to losing the number. Beth must have already known because she handed me a sticky note on which she'd already written down Erica's contact information.

This time I did call, and we agreed to meet at the hospital during Ella's next transfusion. I was so happy to see Ella that I almost cried. She was a beautiful, smart, cheerful little girl. She looked like any "normal" happy kid which made me hopeful for the future of my baby girl. I had so many questions for Ella. I was curious to see if she could feel her port being accessed, what it felt like before and after a blood transfusion, how easy or difficult it was for her to chelate, and the list goes on.

Erica told me about the volunteer work they do with the San Diego Blood Bank thanking donors, and about the Cooley's Anemia Foundation. Everything she shared left me feeling uplifted and inspired. She was so sweet, and told me several times not to hesitate to call if I ever had any questions or just needed someone to talk to. Well, I took her up on it. I called or text whenever a question came up.

A few months later, my husband and I decided to host a blood drive in honor of Kamila's 1st birthday. Again, I can't tell you exactly why- but it took me almost a year after Kamila was born for me to officially "come out" and tell all my friends and family about her condition. I'd told my closest friends and most of my family, but something held me back from full disclosure. I was hosting a blood drive and I wanted it to be successful, so I took that opportunity to tell everyone I knew about her blood disorder, and encouraged all to attend and give blood. Coming out and sharing Kamila's story filled a hole in my heart and made me feel complete. From then on, I felt a strong calling to raise awareness about thalassemia and to advocate for the importance of blood donations

Well, to make a long story longer... I found out that Erica felt that same calling. We partnered up and the following year, hosted a blood drive and fundraiser in honor of World Thalassemia Day 2016. We collected over 100 units of blood for the San Diego Blood Bank and over \$5,000 for the Cooley's Anemia Foundation.

Which brings us to today! Our goal now is to create a support group for local thalassemia patients and families. In all honesty, meeting Erica changed my life. Although I'm pretty sure that Malcolm X wasn't referencing thalassemia with his quote, "When 'i' is replaced by 'we' even illness becomes wellness," I can't think of a truer message when it comes to thriving with thalassemia. This illness CAN become wellness with a strong support system. The inspiration I gained from Erica completely turned around my view of thalassemia for the better, and we want to share this outlook with as many families as we can. We hope to learn from one another and work together to raise awareness within the community. With CAF California Chapter and [thalpalsandiego.com](http://thalpalsandiego.com), we believe we can.



Ella and Kamilla,  
Daughters of Erica  
and Maria

## COOLEY'S ANEMIA FOUNDATION HOSTS PATIENT FOCUSED DRUG DEVELOPMENT MEETING

For the past several months, the Cooley's Anemia Foundation (CAF) has been working diligently to organize a meeting with the U.S. Food and Drug Administration (FDA) to develop an opportunity for patients and caregivers to interact with stakeholders, sharing experiences and stories about the impact of thalassemia on their lives. Though the Foundation's leadership and staff have a long history of working with the FDA and other agencies to support and advocate for patients, July 7, 2017 will be the first chance for all Cooley's Anemia Foundation patients to participate in such a dialogue.

These meetings have the potential to impact future research and development at the FDA and, consequently, the development of new treatments for patients. This Patient-Focused Drug Development meeting ("PFDD") will be open to other stakeholders, and has been scheduled the day before the 2017 Patient and Family Conference. This PFDD process, which was authorized under federal law, has proven beneficial to many groups as they increase their outreach to the FDA. Patients and families will have the option to travel in to the Washington, DC area one day early to attend.

The FDA has expressed interest in learning more about how patients experience thalassemia and the biggest challenges of the disease and of current limited treatment options. The meeting will give patients and families the unique and important opportunity to communicate directly to FDA representatives and to share personal experiences that will be instrumental towards the development of therapeutics to most effectively impact patients. The FDA will be in "listening mode" to learn all they can about the lives of thal patients.

This meeting will be open to the public and provides patients and caregivers with a chance to interact with stakeholders about concerns with current treatment options and hopes for new therapies. Foundation staff will be facilitating the meeting and will be taking notes to use to inform future efforts to improve patient's quality of life and access to therapeutics. The plan is to transcribe the meeting so those who cannot attend may have the opportunity to review the discussion.

The meeting will be split into two major segments and is expected to last approximately five hours. The first topic for discussion will be thalassemia symptoms and the impact of thalassemia on daily life, and the second topic will be patient perspectives and current approaches to treatment. Each topic will include a panel and opportunity for patients to share experiences directly with FDA officials, staff, and other stakeholders.

## THOUSANDS ACROSS COUNTRY JOIN CARE WALK 2017 (CONTINUED FROM COVER)

### CARE WALK 2017 TEAMS:

#teamdailey	Team Colasant
#ZuneeraBrave	TEAM DANIMAC
Acceleron Pharma	Team DNA
Buffalo Cares	Team Emily Grace
Bushkill Rocks!	Team Ercole
ChelationWizards	TEAM FICARRA
Connie's Crew	Team Giuseppe
Corbyn and Cai's Care Walk	Team In Loving Memory of Michelle
Doherty's Team	Team Joshie
Eileen's Team	Team Jovi
Friends of Francesca	Team M&M
Kristy's Thalassemia Warriors	Team Perez
LA Dreambuilders	Team San Diego
Mia's Marchers	Team Staten Island
Mustang Sally	Team Tranquillo
National Distribution Alliance	Team Yanny
Rad's Army	Team Zayna
Saint Luke's Care Walk	Team Zoe
SeaTHALites	Teresa's Carewalk
Siddiqa Dhanji	THE A TEAM
StL Thal Pals	The Little Dinos (In Memory of Andrea Szymanski)
Super Scarlett	Twin Cities Care Walk
Team AJ - Dream for a Cure!	
Team Arizona	

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### FRIENDS OF CAF



# MILANA FAMILY FOUNDATION'S 2<sup>ND</sup> ANNUAL GOLF OUTING FOR CAF WAS A HUGE SUCCESS

***"I would like to thank the Milana Family Foundation from the bottom of my heart for their generosity and the love and compassion they have shown our patients as a major donor of CAF"***

– Anthony Viola, CAF National President

The 2nd Annual Golf Outing and Poker Tournament held by the Milana Family Foundation on June 12 at the Fresh Meadow Country Club in Lake Success, NY was a tremendous success. The Cooley's Anemia Foundation and Stop Abuse, a nonprofit fighting child abuse, will receive the proceeds from this event, thanks to the generosity of Tom Milana and the Milana Family Foundation. Last year, CAF and Stop Abuse each received \$75,000 from the proceeds of the 1st Annual Golf Outing and Poker Tournament.

"I would like to thank the Milana Family Foundation from the bottom of my heart for their generosity and the love and compassion they have shown our patients as a major donor of CAF," said Anthony Viola, CAF National President.

"At the dinner that night, I was introduced to a Cooley's patient who just got married a couple of weeks before this golf outing, by Adriana Milanam," continued Mr. Viola. "The patient had kept her story silent for years but it appears like she will start working with CAF to make her successful story known to other patients and to help CAF any way that she can. This was one of the highlights of the day, and another example of how CAF is reaching out to more and more patients throughout the United States. A great day for CAF, their patients, and their families, for sure."

Mr. Viola encourages the thalassemia community to reach out and show gratitude to the Milana family, saying "as a proud Foundation, please feel free to share your gratitude and appreciation to the Milana Family Foundation. And thank you, again, to the Milana Family."

"It is very inspiring to see how dedicated people are to this fight against thalassemia," said Thomas Milana. "That made our desire for a successful event even greater. Although my family is not directly affected by thalassemia, we are very fortunate and feel absolutely honored to have worked with the Cooley's Anemia Foundation."



## Being adherent to thalassemia treatment has a big payoff – a longer life.

**Studies have shown that people with thalassemia who follow the recommended treatment plans for thalassemia have better health outcomes than people who don't.**

**So even if you can't tell that your treatment is making a difference, your body can!**

Sticking with transfusions, chelation and other treatments can be difficult – but it's worth it.



This message was developed as part of Cooperative Agreement #1U27DD001150-01 from the Centers for Disease Control and Prevention.

## CAF AWARDS \$260,000 FUNDING IN MEDICAL RESEARCH FELLOWSHIPS (CONTINUED FROM PAGE 3)

formation of HbF and theoretically would provide additional benefit for patients undergoing gene editing therapy. In the second approach, she aims to develop gene therapy constructs with the use of non-integrating adenoviral vectors, to facilitate an efficient delivery to patient. The significance to thalassemia is high. Only a third of patients have a suitable donor and approaches to modify stem cells are a possible way to a cure.

**Antonella Nai, PhD**, of San Raffaele University in Milan, Italy, will be examining whether TfR2 (a gene mutated in the iron overload disorder, hereditary hemochromatosis) expression in red blood cells of wild type and thalassemic mice can increase red blood cell number in the short term. Dr. Nai will use a number of approaches to inhibit TfR2 activity in the developing red blood cells of a thalassemic mouse model through her study, "Transferrin Receptor 2 : A Novel Potential Therapeutic Target for B-Thalassemia." In collaboration with Ionis Pharmaceuticals, Inc., Dr. Nai will test the use of antisense oligonucleotides (ASOs), to reduce TfR2 expression in developing red blood cells. Prior work from Drs. Nai and her mentor, Dr. Clara Camaschella, using the genetic approach of deleting TMPRSS6 in thalassemic models is ready for phase I clinical trials, demonstrating the success of this team in the identification and validation of clinically relevant targets for thalassemia.

**Lei Yu, PhD**, of University of Michigan Medical School, will be focusing on development of a small molecule analog to inhibit the activity of a repressor complex of fetal globin expression through his study, "Development of RN-1 Analogs to Inhibit LSD1 Activity as a Strategy to Treat B-Thalassemia." This study builds on fine-tuning a molecule that has already shown high promise in pre-clinical models; hence it is already closer to clinical application and thus merits the efforts to optimize its properties. Appropriate, in-house collaborations on pharmacology and efficacy have also been set up and thus increase confidence in the ultimate success of the approach.

**Daniel Lucas, MD, PhD**, of University of Michigan Medical School, will conduct a set of studies proposed to improve hematopoietic reconstitution (recovery of the bone marrow and its environment) after hematopoietic stem cell transplantation. Preliminary data from both well-controlled mouse experiments and retrospective analysis of data from human thalassemia patients who have undergone transplant suggests that time to marrow recovery and avoidance of "graft failure" can be improved as the number of bone marrow-derived granulocytes increases. Dr. Lucas's study, "Identification of the cellular targets through which granulocytes drive hematopoietic regeneration after transplant," is well suited to determine both the role of TNF-alpha in this process and its potential cellular target in the perivascular niche. This work has tremendous potential to help advance hematopoietic stem cell transplantation, particularly when limited donor cells are available.

**Julia Xu, MD**, of Duke University, aims to understand understand the beliefs, attitudes, and behaviors of Myanmese and Lao migrant populations towards thalassemia in her study, "Thalassemia Screening in Myanmese and Lao Migrants in Thailand." Little is currently known about perceptions of blood disorders, inherited diseases, and prenatal testing in these vulnerable populations where thalassemia is highly prevalent. The information obtained from these studies can be used to develop thalassemia screening and prevention programs, targeted education, and health care programs for this population. Given the large burden of thalassemia in Thailand and Southeast Asia and changing migration patterns, with increasing numbers of immigrants from these regions to the United States, the knowledge gained from this grant should have a significant global impact, aiding in the development of culturally-appropriate screening programs and health care delivery.

### RENEWAL FELLOWSHIP RECIPIENTS

**Katie Carlberg, MD**, of the Children's Hospital Oakland, is developing a noninvasive approach to prenatal diagnosis of thalassemia in her study, "SNP Discovery and Characterization of the Human Beta-Globin Gene for Non-Invasive Prenatal Testing for Beta-Hemoglobinopathies." Current approaches to prenatal diagnosis of thalassemia are invasive and increase risk of morbidity to mother and fetus. This noninvasive approach is based on recent advances in DNA isolation and enrichment and DNA sequencing, and could be accomplished as early as 8 weeks of gestation. Dr. Carlberg has made excellent progress in Year 1 and in Year 2 she will carrying out rigorous testing of this approach in a well-defined thalassemia population. Dr. Carlberg's research will establish a critical experimental foundation to developing a cutting-edge major advance in this field.

"Thalassemias are some of the most common autosomal recessive (AR) disorders in the world," Dr. Carlberg explains. "The development of non-invasive prenatal testing (NIPT) for AR disorders has proven challenging as it requires knowing which allele was transmitted to the fetus from each parent... We will investigate an indirect method that involves calculating the ratio of sequence reads for the two maternal beta-globin alleles and comparing this ratio from the maternal plasma DNA to the maternal whole blood DNA."

**Merlin Nithya Gnanapragasam, MD, PhD**, of Icahn School of Medicine at Mount Sinai in New York City, is performing experiments in HuDEP-2 cells to examine the consequences of mutation of the EKLF upstream enhancer region in her project, "Genome Editing of EKLF Enhancer Elements for Fetal Hemoglobin Induction." She will also be experimenting with homology directed recombination of JMML intron enhancer mutation into human erythroid progenitor HuDEP-2 cells to test the effect on erythroid differentiation and globin gene expression.

"Beta thalassemia is one of the most common inherited disorders," Dr. Gnanapragasam explains. "An attractive therapeutic strategy to ameliorate and potentially cure beta thalassemia is to increase fetal hemoglobin levels in the red blood cells of people who have thalassemia. Erythroid Kruppel Like Factor (EKLF/KLF1) is a transcription factor that is essential for the proper regulation of hemoglobin genes. My goal is to identify strategies to in-

crease fetal hemoglobin levels, by modulating EKLF's regulation of globin gene expression using genome editing."

Karen Finberg, MD, of Yale Medical School, is studying the process of how the gene NCOA4 mediates the degradation of ferritin in her study, "The Role of NCOA4 in the Regulation of Hepatic Iron Stores." She will be using mouse strains exhibiting different extremes of iron regulation to conduct her experiments. Her findings could prove that increasing NCOA4 could accelerate the removal of excess iron from the liver, which then would be removed by chelation therapy. In the first year she has completed the first phase successfully and she proposes to next determine how this protein acts in the liver to regulate iron homeostasis in mice.

"How iron is released from the body's storage sites... is currently very poorly understood," says Dr. Finberg. "In this study, we plan to use mouse models and cultured cells to investigate the biological processes that enable iron to be removed from the liver, the body's major organ of iron storage. It is our hope that our research findings will contribute to improved clinical outcomes for patients with β-thalassemia by providing knowledge that will aid in the development of new therapeutic approaches to promote iron removal from the body."

**With thalassemia, you can't afford to wait for your heart to tell you it has too much iron.**

**Iron can start building up in the heart for a long time before a person with thalassemia feels any effects. But getting iron out of the heart as early as possible is important for the best health outcome.**



**That's why people with thalassemia need to have regular T2\* cardiac scans performed, so that their doctors can take steps to keep their hearts healthy.**

**Noninvasive testing to see if iron is in the heart is the best way to know if your chelation therapy is keeping your heart healthy.**

This message was developed as part of Cooperative Agreement #1U27DD001150-01 from the Centers for Disease Control and Prevention.



## WITH YOUR HELP, THE CURE IS WITHIN REACH!



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## THOUSANDS ACROSS COUNTRY JOIN CARE WALK 2017



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