

# CONNECTIVE ISSUES

WINTER 2017

**KNOW THE SIGNS.  
FIGHT FOR VICTORY.**



THE **MARFAN**  
FOUNDATION



**The Marfan Foundation creates a brighter future for everyone affected by Marfan syndrome and related disorders. We work tirelessly to advance research, serve as a resource for families and healthcare providers, and raise public awareness. We will not rest until we have achieved victory.**

**Learn more and get involved at [Marfan.org](http://Marfan.org).**

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## CONNECTIVE ISSUES

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THE MARFAN FOUNDATION  
22 MANHASSET AVENUE  
PORT WASHINGTON, NY 11050  
516-883-8712 | 800-8-MARFAN  
[WWW.MARFAN.ORG](http://WWW.MARFAN.ORG)

**COVER:** TAHLULAH (LULU) BUTVICK, 2, OF GARDEN CITY, NY, WITH HER MOTHER, SARA MAHER. BECAUSE LULU WAS DIAGNOSED WITH MARFAN SYNDROME EARLY, HER CONDITION IS BEING MANAGED SO THAT SHE CAN ENJOY A LONG LIFE.

## HAPPY NEW YEAR!



Although we each come to The Marfan Foundation through our own path and are on our own journey, we share a common bond and commitment to creating a brighter future for everyone affected by Marfan syndrome and related disorders—a bond and a commitment that helps to enhance and save lives every day.

As the President and CEO of The Marfan Foundation, I wake up every morning with a goal of doing my part to help move our mission forward. None of us have a “casual” relationship with Marfan or related disorders and it is our shared commitment and combined strength that is making a difference. Our success is built on the shoulders of each other, and every single volunteer, staff member, physician, scientist, and donor wears a Marfan jersey.

I also believe that there is no organization that works harder at being a steward of the donated dollar. We do our best to make certain that every gift goes toward accomplishing our mission. EVERY dollar truly matters at The Marfan Foundation.

My wishes for the New Year:

1. The number of regional symposiums continues to increase, we touch people in a meaningful way, and introduce them to the Foundation and individuals on a similar medical journey.
2. We continue to fund the most innovative research because we know somewhere, something incredible is waiting to be known. Thank you, Dr. Carl Sagan!
3. While we enjoy great success in generating new financial support, our “Donations in Memory of” list gets smaller and smaller. This past year, we lost way too many friends and loved ones to Marfan syndrome and related disorders.
4. We have a record year in people contacting our Help and Resource Center. It is an amazing resource!
5. We have the most successful, spirited annual conference in our history in Atlanta! Thank you, Emory University and Sibley Heart Center. We could not be more excited!

In closing, I would like to dedicate this column to my friend and our longtime supporter, Rita Eisman. We lost Rita in November, after she and her husband, Sandy, attended 30 consecutive conferences. Rita clearly has a seat in the Marfan Hall of Fame. We miss you!

Regardless of how you may be involved in The Marfan Foundation, remember Milton Berle’s quote, “If opportunity doesn’t knock, build a door!”

Victory Is . . .

Michael L. Weamer  
President & CEO



SANDY AND RITA EISMAN

# REGIONAL SYMPOSIUMS

Creating connections in Houston and Fort Lauderdale



JOE DIMAGGIO CHILDREN'S HOSPITAL HOSTED THE FLORIDA SYMPOSIUM, WITH GENE DX PROVIDING CRUCIAL SUPPORT. PEOPLE WITH MARFAN AND RELATED DISORDERS AND THEIR FAMILY MEMBERS CAME FROM ALL PARTS OF THE STATE TO LEARN AND TO GET INVOLVED WITH THE MARFAN COMMUNITY. THE KEYNOTE SPEAKER WAS DR. JUAN BOWEN, DIRECTOR OF THE MARFAN CLINIC AT THE MAYO CLINIC IN ROCHESTER, MN. HE IS PICTURED HERE WITH BERT MEDINA, A MEMBER OF THE FOUNDATION'S BOARD OF DIRECTORS, AND DR. ROSHA MCCOY, CHIEF MEDICAL OFFICER OF JOE DIMAGGIO CHILDREN'S HOSPITAL.

“Knowledge is power, so I couldn't pass up the chance to learn more about treatments, testing, and advancements—from both expert medical providers and fellow Marfs. . . . It was also a chance to meet other members of our Florida community in person who I had only met before online, and ideally connect with others from the Central Florida area to help expand our local group—which I did.”

– DAWN OCKSTADT

Our expanding regional symposium program continues to bring one-day conferences to cities around the country so that people with Marfan syndrome and related disorders can get up-to-date medical information from local experts and meet people who are on the same medical journey. In December, we held symposiums in Houston and Fort Lauderdale. Upcoming symposiums are in Nashville (March 11), Boston (June 8), and Cleveland (date TBD).



Every symposium offers a separate program for children and teens so they can meet others who have the same diagnosis. These children did activities together at the Houston event.



The Houston symposium, sponsored by Invitae, had an impressive line-up of speakers, led by our Professional Advisory Board member Dr. Dianna Milewicz, President George H.W. Bush Chair of Cardiovascular Medicine and director of the Division of Medical Genetics of UT Health (seen here speaking with members of the Marfan community).



ROSELINE ROSARIO (L), WHO HAS LOEYS-DIETZ SYNDROME, AND DAWN OCKSTADT, WHO HAS MARFAN, ARE BOTH FROM ORLANDO, BUT MET FOR THE FIRST TIME AT THE FLORIDA SYMPOSIUM.

# 2016 RESEARCH GRANTS

\$1.2 million awarded for seven new research initiatives



2016 FACULTY GRANT RECIPIENT JESSICA WAGENSEIL, DSC, WASHINGTON UNIVERSITY

Each year, The Marfan Foundation pursues the most innovative research and makes sure that it receives proper funding. In 2016, we received 22 grant proposals and awarded seven new grants to support promising research by scientists and physicians. Through our grant program and other commitments, we funded \$1.2 million in research this year.

As a result of our efforts, grants will help researchers:

- Investigate therapies for mitral valve prolapse, a leading cause of mortality in infants and children diagnosed with Marfan syndrome before age 4
- Study whether or not regular exercise improves aortic health
- Explore coping skills in adolescents with Marfan syndrome
- Identify blood biomarkers to monitor aortic root size
- Test new therapeutic options on aneurysm growth
- Develop a new animal model of aortic aneurysm using zebrafish
- Investigate the relationship between abnormally shaped arteries, the size of aneurysms, and their likelihood of dissecting

- Look into the role of proteins in Beals syndrome as well as in the muscle weakness and loose joints seen in Marfan syndrome

## 2016 Research Grant Recipients:

Maryline Abrial, PhD, Massachusetts General Hospital  
*Investigating a Novel Genetic Model of Aortic Root Aneurysm in Zebrafish*, \$100,000 2-year McKusick Fellowship Grant

Vallabhajosyula Prashanth, MD, MS, University of Pennsylvania  
*Plasma Endothelial Specific Exosome Profiling in Marfan Syndrome*, \$75,000 2-year Early Investigator Grant

Rouf Rosanne, MD, Johns Hopkins School of Medicine  
*Using an Established Marfan Syndrome Mouse Model to Understand Mitral Valve*

*Prolapse Pathogenesis*, \$75,000 2-year Early Investigator Grant

Dr. Suneel Apte, MBBS, DPhil, Cleveland Clinic  
*Fibrillin Microfibril Regulation by ADAMTSL3*, \$100,000 2-year Faculty Grant

Seda Tierney, MD, Stanford University, and Mitra Esfandiarei, PhD, Northwestern University  
*Children and Adolescents with Marfan Syndrome: 10,000 Healthy Steps and Beyond*, \$100,000 2-year Faculty Grant

Zhiyong Lin, PhD, Case Western Reserve University  
*Allosteric Activation of PP2A to Limit the Progression of Aortic Aneurysm in Marfan Syndrome*, \$100,000 2-year Faculty Grant

Jessica Wagenseil, DSc, Washington University  
*Arterial Tortuosity and Aneurysms*, \$100,000 2-year Faculty Grant

If you would like to participate in research on Marfan syndrome and related disorders, please visit:  
[www.marfan.org/current-studies](http://www.marfan.org/current-studies)

# MEET YOUR GENE

An Introduction to the Marfan Gene and Current Research: A conversation with Hal Dietz, MD

*Roanne Weisman, a healthcare writer from Boston who has Marfan syndrome and who is long-time member of the Foundation, had a conversation with Hal Dietz, MD, Victor A. McKusick Professor of Medicine and Genetics, and Director, William S. Smilow Center for Marfan Syndrome Research, Johns Hopkins University School of Medicine, to get a basic understanding of Marfan research to share with our community. Here are excerpts. The entire Q & A is on The Marfan Blog.*

## What is the Marfan gene and how does it cause Marfan syndrome?

The Marfan gene encodes for fibrillin-1, which is a component of connective tissue—the material between the cells of the body that binds cells together and gives tissues form and strength. Besides acting like “glue,” connective tissue proteins such as fibrillin-1 can provide instructions to neighboring cells that influence how they behave. There is strong evidence that fibrillin-1 participates in both types of functions. As with most other genes, every person normally has two copies of the fibrillin-1 gene. In people with Marfan syndrome, at least one of these copies has a defective sequence of DNA—also called a mutation—so the “instructions” to make fibrillin-1 are not quite right. As a result, the altered fibrillin-1 has a reduced ability to perform its intended structural and instructive functions.

## What is the focus of current research to improve the lives of people with Marfan syndrome?

Thanks to years of research, we came to the understanding that the fibrillin-1 protein also serves other important functions besides structural functions, and the course of our therapeutic focus has become much more promising.

Currently, there is a strong research focus on the ability of fibrillin-1 to regulate the activity of a class of molecules, called growth factors. These molecules bind to the surface of cells and tell the cells how to behave. One growth factor, TGF-beta, has particular relevance for Marfan syndrome. Normally, TGF-beta is active during fetal development, encouraging growth, but is less active in adults, except at certain times, such as for wound healing. Normally functioning fibrillin is like a “traffic cop” for TGF-beta, signaling it to be active when it is needed and stopping or suppressing its activity when it is not needed. But in people with Marfan syndrome, this signaling system has trouble stopping the activity of TGF-beta. We learned that in both humans and mouse models with Marfan syndrome, there was clear evidence for too much TGF-beta activity.

## What does too much TGF-beta activity do to the Marfan body?

We and others have focused on problems with the aortic wall enlarging and dissecting, but there is also good work that shows high TGF-beta activity contributes to problems in heart valves, skeletal muscles, and lungs.

## Has this knowledge led to a therapeutic approach (medication)?

Recent studies have suggested that medications such as losartan show strong promise for the care of people with Marfan syndrome and related disorders—performing as good as or better than conventional therapies, such as beta blockers, in various studies. There is both room for improvement and many unanswered questions. Is losartan the best drug in its class (a group of medications called angiotensin receptor blockers or ARBs)? What is the optimal dose? Are there some people who will respond to ARBs and others who will not? Are there combination therapies that should be considered? Answers to these critical questions require more research and future clinical trials.

## Is personalized medicine on the horizon for people with Marfan syndrome?

Individualized treatment is one of the most exciting and promising Marfan treatment research pathways right now. One day we may be able to design the treatment that would be right for each person. That is the goal of the ancillary studies that are still ongoing. We hope that we will be able to look at the genetic makeup of each person, as well as specific measurements of chemicals in the bloodstream, to determine how people are responding to treatments. This will help us predict whether we are on the right track or if we need to modify treatment.



HAL DIETZ, MD. PHOTO BY MATT ROTH / MATTROTHPHOTO.COM



# WALK FOR VICTORY



## UPCOMING WALKS

Phoenix, AZ	March 5
Houston, TX	March 25
Cleveland, OH	March 26
Atlanta, GA	April 29
Pasadena, CA	May 20
New York, NY	May
Boston, MA	June

Walk for Victory is our nationwide walk program held in various cities across the country to bring the Marfan syndrome and related disorders community together. The walks are non-athletic, family-oriented events focused on fundraising and fun. Walks in 2015-2016 raised more than \$300,000 for the Foundation's programs and services, which reach more than 100,000 affected individuals, families, and physicians each year. In our 2016-2017 season, we hope to raise more than \$500,000 to support quality of life programs for our families, kids, teens, young adults, and seniors living with Marfan and related disorders. These walks provide a great opportunity for the community to come together to raise critical funds for the Foundation.

Interested in joining us at one of our upcoming walks? Go to [Marfan.org/Walk](http://Marfan.org/Walk) to register.

If you have questions or want to learn more, contact Kim Huddleston, our national walk director, at [khuddleston@marfan.org](mailto:khuddleston@marfan.org).

# PROFESSIONAL ADVISORY BOARD NEWS

Heidy Connolly, MD, Mayo Clinic, is new chair



### New Chair

The Marfan Foundation is pleased to announce that Heidi Connolly, MD, is now the chair of its Professional Advisory Board. This board, which is comprised of the leading Marfan syndrome and related disorders clinicians and researchers from all over the U.S., provides the Foundation with professional expertise and oversight of all medical informa-

tion on the diagnosis and management of Marfan syndrome and related disorders. In addition, these doctors advise us on research opportunities and priorities.

Dr. Connolly, who has served on the Professional Advisory Board since 2006, is a Professor of Medicine at the Mayo Clinic College of Medicine in Rochester, MN, and a Consultant in Cardiovascular Diseases at the Mayo Clinic. In addition, she is director of the Mayo Clinic's Congenital Heart Center and Adult Congenital Heart Disease Clinic, serves as Vice-Chair for Education in the Department of Cardiovascular Diseases at Mayo Clinic, and is an active member of the Echocardiography Laboratory. Dr. Connolly established the Marfan Syndrome and Thoracic Aorta Clinic at Mayo Clinic and served as its director until 2012. She played a leading role in our 2009 and 2016 annual conferences, which were hosted by Mayo Clinic.

Dr. Connolly replaces Dr. Alan Braverman as Chair. Dr. Braverman remains as a valued member on the Professional Advisory Board.



### New Member

Leonard N. Girardi, MD, Professor of Cardiothoracic Surgery in the Department of Cardiothoracic Surgery at Weill Cornell Medicine, has been named to our Professional Advisory Board.

In addition to his clinical care, Dr. Girardi is committed to training the next generation of cardiothoracic surgeons. He is a member of numerous surgical societies and has published extensively on multiple topics in cardiovascular and aneurysm surgery and has given many lectures at regional and national surgical meetings.

In 2004, Dr. Girardi received the Hero with a Heart Award from The Marfan Foundation for his surgical expertise in the care of patients with Marfan syndrome and related disorders. In 2016, he was honored again at the Foundation's Heartworks gala in New York as part of the Weill Cornell Medicine cardiac care team, along with Dr. Richard Devereux and Dr. Mary Roman. Dr. Girardi has also been honored with the Stephen Gold Award for Humanitarianism in Medicine, Il Leone di San Marco Award in Medicine, Coryell Prize in Internal Medicine, and more.



### On The Move

Dr. Duke Cameron, formerly the Cardiac Surgeon-in-Charge and James T. Dresher Sr. Professor of Surgery at Johns Hopkins Hospital, moves to Massachusetts General Hospital on February 1 as Co-Director of the Thoracic Aortic Center and Surgical Director of the Adult Congenital Heart Disease Program.

### Managing Your Condition

Our Professional Advisory Board plays a critical role as they provide oversight to all the medical information we share with our community on Marfan syndrome and related disorders. This includes fact sheets on how these conditions are diagnosed and how various features are treated—heart and blood vessels, bones and joints, eye, lungs, skin, nervous system, and teeth. There are also fact sheets on conditions related to Marfan syndrome, such as Loey's Dietz syndrome and Ehlers Danlos syndrome.

The fact sheets can be downloaded for free at our website, Marfan.org. Just look for patient resources. Several of the fact sheets are also available in Spanish.



## EMERGENCY PREPAREDNESS

Following your doctor’s recommendations for medication, monitoring, and physical activity gives you the best chance of avoiding a serious complication of Marfan syndrome or a related disorder. Still, there are no guarantees. That’s why it’s important for you to learn about the medical problems that could arise and require immediate medical treatment.

The most serious of these problems involve the heart and blood vessels, but there are other problems involving the eyes and lungs that also need emergency treatment.

To help prepare you for these situations, we recommend that you complete our Emergency Preparedness Kit, which we created specifically for people with Marfan syndrome and related disorders. It includes our Emergency Alert Card which you can download and carry in your wallet. These are available in the “patient resources” section of our website, Marfan.org.



Hospital emergency departments are also accustomed to looking for emergency alert bracelets. There are many different styles. Check with your physician to determine the appropriate wording for your bracelet.

## HELP & RESOURCE CENTER: HERE FOR YOU

More than 2,000 people contact the Foundation’s Help & Resource Center every year. Our nurse, Amy Kaplan, RN, BSN, notes that she receives an array of questions that include general and specific medical questions; psychosocial questions; and insurance inquiries.

Individuals often call with diagnostic questions, asking about the evaluation process, specific physical features, genetic testing findings, and disorders related to Marfan syndrome.

Currently, the five areas that people most frequently contact the Help & Resource Center about are:

- Physical activity guidelines
- Medical specialists (and referrals)

- Managing symptoms, including pain
- Managing aortic problems, including medications and surgery
- Gastrointestinal issues, including colonoscopy

“In every case, the information we provide is general in nature, as we don’t have anyone’s specific medical situation,” said Amy. “I encourage people to take our information and discuss it with their personal physician.”

If you have questions about Marfan syndrome or a related disorder, please call 516-883-8712, ext. 126, or email [akaplan@marfan.org](mailto:akaplan@marfan.org).

## WAYS TO GIVE

In this issue of *Connective Issues*, you are reading about our research program, Help & Resource Center, regional symposiums, annual conference, awareness efforts, new Marfan camp for families, and emergency medicine campaign. These programs require ongoing support from people like you. Your donations—large and small—enable us to create a brighter future for everyone living with Marfan syndrome and related disorders.

Here are some ways you can directly impact the lives of people in our community.

- Become a monthly donor by committing a certain amount of money each month. You can make a difference for as little as \$10 monthly!
- Make a tribute donation in honor, or in memory, of a loved one.
- Donate through a will or bequest, life insurance, stock, appreciated property, charitable remainder trust, or charitable lead trust.



Questions? Please contact Helaine Baruch, chief development officer, at [hbaruch@marfan.org](mailto:hbaruch@marfan.org), or 516-883-8712, ext. 141.



# BEFORE YOU START TO EXERCISE

## Guidelines for your conversation with your doctor

**Medical experts recommend** that, if you or your child has Marfan syndrome or a related disorder, you talk with your doctor before pursuing any exercise regimen or playing a sport (or continuing to play a sport). While you are most interested in knowing if you or your child can *[fill in the blank with your activity]*, there are related issues to discuss with your doctor. As a result, you will understand what is safe, what isn't safe, and why.

Dr. Alan Braverman, director of the Marfan Clinic at Washington University in St. Louis, and Dr. Ron Lacro, director of the Cardiovascular Genetics Clinic at Boston Children's Hospital, suggest asking the following questions.

- What types of physical activity are safe for me?
- Why do I (or my child) need to modify exercise levels?
- Because of the way Marfan (or a related disorder) affects me, how will this impact my ability to exercise or do physical activity? Note that this could be related to eyesight, orthopedic issues (involving feet, hips, back), or lungs/thoracic issues.
- Are there certain types of exercise that I should avoid? Why?
- What about doing exercise that involves competition? What levels are safe?
- Can I lift weights? If so, how much? Note that the general guidelines for safe weightlifting in Marfan syndrome involve lifting very low amounts of weight with repetitions that does not reach straining or muscle fatigue. Check with your personal physician about specific conditions that are safe for you.



CARSON FLANAGAN, 12, OF WACO, TX, HAS MARFAN AND HAS BEEN PLAYING GOLF SINCE HE WAS 4.

“Not everyone is the same. For some with Marfan syndrome or a related disorder, the major issue is eyesight or the skeleton while, for others, it is the aorta,” said Dr. Braverman. “It is important to remember that low to moderate levels of aerobic exercise to a level that does not involve straining is typically very safe for most people.”

In general, the focus of exercise should be fun and fitness, rather than competition. And while some school or community sports leagues may be permissible in younger children, restrictions may be needed in the teen years as the competition increases. In anticipation of sports limitations, it may be beneficial for parents to encourage diverse interests that encompass a variety of sports as well as non-sports activities.

## PHYSICAL ACTIVITY GUIDELINES

Physical activity is important to everyone. As always, we encourage people with Marfan and related disorders to consult with their doctors prior to beginning any exercise regimen to ensure they are participating at a level that is safe for them.

The Foundation's Physical Activity Guidelines as set forth by its Professional Advisory Board, comprised of the world's leading medical experts on Marfan syndrome, remain unchanged. These guidelines say that:

*In general, most people living with Marfan syndrome should exercise regularly through low-intensity, low-impact activities adapted to meet their specific needs. They should avoid contact sports because of the risk of damaging the aorta and injuring the eyes. Strenuous activities, such as competitive sports and weightlifting, also should be avoided because of the stress placed on the aorta.*

Dr. Alan Braverman, an adult cardiologist, and Dr. Ron Lacro, a pediatric cardiologist, who are both on our Professional Advisory Board, discussed physical activity and Marfan and related disorders on a recent conference call. The recording is available on the Foundation's website.

If you have general questions about exercise and Marfan or a related disorder, contact our Help & Resource Center at [support@marfan.org](mailto:support@marfan.org). For specific questions, contact your physician who is familiar with your personal medical situation.

## MARFAN FAMILY CAMP WEEKEND IN APRIL

If your child with Marfan or a related disorder wants to go to camp, but you are reluctant to let them go alone, here's an opportunity for you and your family. For the first time, The Marfan Foundation is hosting a family camp weekend in an environment that is fun for the whole family and safe for your child with Marfan or a related disorder.

Our family camp weekend is April 21-23, 2017, at the beautiful Twin Lakes campgrounds in Winder, GA, just 40 minutes outside of Atlanta. The camp is fully accessible and will have a Marfan-knowledgeable nurse on premises.

There will be special activities planned for families and children of all ages, and separate activity times for parents and children. The program includes arts and crafts, camp fires, music, gym, fishing, boating, hiking, and more. Parents will also have an opportunity to have professionally led discussions with other parents, as well as informal time for creating connections.

The cost to attend is \$80 per adult and \$50 per child (up to the age of 12). This includes accommodations (cabins with beds, showers, toilets) and all meals.

To register, visit [Marfan.org](http://Marfan.org). If you have questions or would like additional information, please contact Susan Leshen, senior director of patient and program services and volunteer leadership, at [sleshen@marfan.org](mailto:sleshen@marfan.org) or 516-883-8712, ext. 122.



## HOW GREAT LIFE BECOMES

When you finally accept that you have Marfan syndrome

By Samantha Malone

A year ago, I was different then I am today. A year ago I was still in my thirties, a wife, and a mother.

A year ago, I was hiding something. I was ashamed to disclose a very important piece of my life. I had an unrealistic fear of letting this secret out.

Marfan syndrome was that secret.

I was diagnosed at the tender age of two. I didn't start out life keeping this secret. I told anyone, and was never ashamed. That changed as I got older. I started to understand my condition. I understood that I would live my life differently. I was different. That was a hard pill to swallow. I heard the ugly words from schoolyard bullies. I saw the look of pity on others' faces when I told them my story.

I decided to lock away my Marfan syndrome and only discuss it with my family and doctors.

I carried the weight of the shame every day. It was a weight I put on myself that wasn't necessary.

I was finally OK with talking about my disorder. It has been one of the most freeing things I have ever done.

This past summer, on a gorgeous July evening, I was sitting on my back porch scrolling through my Facebook feed. I stumbled upon a site that posted blogs from people living with disabilities and illnesses. At the end of each blog post, there was a link on how to submit your own story. Like dominos tumbling over each other in a perfect pattern, things started falling into place in my mind. I started typing and wrote my very first blog post. It was about being a mother with Marfan syndrome.



SAMANTHA MALONE IS A BUSY MOTHER TO TWO AMAZING LITTLE BOYS WHO IS MARRIED TO HER HIGH SCHOOL SWEETHEART. WRITING FROM HER HOME IN A SMALL TOWN IN OHIO, SHE HAS LEARNED HOW TRULY IMPORTANT IT IS TO APPRECIATE THE BEAUTY IN EVERY MOMENT THAT SHE IS GIVEN.

Two weeks later, the post was published. It was the moment I let my secret out. I updated my status on social media with the link to my blog. I was coming out of the Marfan closet to the whole world in the form of a public blog post on a social media platform. There was no looking back.

You want to know what happened when I let my secret out? The only thing that changed was me. People treated me the same; nothing negative happened. I was finally OK with talking about my disorder. It has been one of the most freeing things I have ever done. I broke the chains of fear and shame. I don't have this secret gnawing away at me. I can freely talk about my health without becoming uncomfortable.

I understand how uneasy you may be to open up about Marfan syndrome, but I can tell you that the pride you have once you've opened up about it is an amazing feeling. Be free from your fear because you have nothing to fear. I know.

# FEBRUARY IS MARFAN AWARENESS MONTH

More awareness still needed for early diagnosis and emergency situations

Eleven years ago, at the age of 29, Yanira Dimas, of California, went to a hospital emergency department with various symptoms, including chest pain. She was quickly diagnosed with a severe migraine and released. Two days later, still experiencing the same symptoms, she went to another hospital emergency department. This time, through imaging, the doctors saw her enlarged aorta and she was transported to a teaching hospital where she had aortic surgery. After her recovery, Yanira had genetic testing and the Marfan diagnosis was confirmed.

“Sometimes, we trust the doctors because we have this notion that they know what’s best since they studied medicine for so long,” said Yanira. But, as she learned, this is not always the case. Many people are still not diagnosed with Marfan until an emergency arises.

During Marfan Awareness Month (February), it is critical to raise awareness of Marfan and related disorders so that people are diagnosed early before a potentially fatal aortic dissection. “Know the signs” rings true both for early diagnosis as well as for recognizing aortic dissection.

People with Marfan syndrome can live a normal lifespan, but they must be diagnosed and treated. While significant strides have been made in the awareness of Marfan syndrome and related disorders, there is much more to be done.

**During Marfan Awareness Month, take action!** Share information on social media, tell your story to the local newspaper, bring information to doctors’ offices, or arrange for an informational table at a local mall, library, or school. Or come up with another way to put Marfan and related disorders in the public eye in your city. We are here to help. If you need assistance, please contact us at [volunteer@marfan.org](mailto:volunteer@marfan.org).



**TOP:** YANIRA DIMAS, WHO WAS DIAGNOSED AFTER AN AORTIC DISSECTION

**BOTTOM:** TAHLULAH (LULU) BUTVICK, DIAGNOSED AT 20 MONTHS OF AGE, IS ALREADY UNDER A DOCTOR’S CARE. HER PROGNOSIS IS BRIGHT BECAUSE OF THE EARLY DIAGNOSIS AND EARLY TREATMENT.



**CONNECT  
WITH US**



on social media and raise awareness  
of Marfan syndrome and related disorders in  
February and throughout the year!



**YouTube**



# OUR COMMUNITY MAKES A DIFFERENCE NATIONWIDE



KYLER ROMANO (STANDING) WITH HIS BROTHER, KADEN

The Marfan and related disorders community—young and old, from coast to coast—continues to show its dedication to creating a brighter future for everyone living with these conditions. Awareness and fundraising events—regardless of how large or small they are—keep Marfan and related disorders in the public eye. The awareness leads to more people getting an early diagnosis while the funds raised support the Foundation’s programs and services (e.g., annual conference, regional symposiums, Help & Resource Center, and so much more!).

The Romano family, of Rhode Island, has been involved in fundraising and awareness since Kyler, now 11, was diagnosed in 2012. They raised more than \$7,000 through a youth baseball tournament and dad, Jay, is going to teach a self-defense course for women in April, with registration fees going to the Foundation. Jay says that “getting Kyler involved with raising money and awareness makes him more comfortable with the diagnosis.”

Liam Nelson, a member of the Foundation’s Teen Council who is from Atlanta, launched a fundraising and awareness campaign in November 2016. The public awareness components of the campaign include presenting to local high school sports coaches about Marfan syndrome and doing presentations to students in his school. As far as the

fundraising is concerned, Liam, who is halfway towards his \$1,000 goal, has “experienced first-hand the overwhelming positive impact made in the lives of those with Marfan syndrome” as a result of the Foundation’s work; that’s why he’s motivated to give back.

Among the longest running community events is the casino night organized by Michele Cripps in California every April. Over the past 12 years, the casino night has raised more than \$67,000. Originally a joint project with the local Tall Club, casino night is both a successful fundraiser and a really fun night out.

“Casino night is important to me because it shows those with Marfan it is okay to tell other people that you have Marfan and they will support you,” said Michele. “We can do anything we put our minds to, everyone has fun, AND it raises money.”

The next casino night is April 29, 2017.

Mark your calendar and check our website for more details if you would like to attend.

It’s easy to set up your own fundraiser! Simply go to [Marfan.org/CommunityFundraisers](http://Marfan.org/CommunityFundraisers). For assistance, please contact us at [volunteer@marfan.org](mailto:volunteer@marfan.org).



MICHELE CRIPPS (RIGHT) BEGAN ORGANIZING A CASINO NIGHT IN CALIFORNIA TWELVE YEARS AGO

# ANNUAL CONFERENCE COMING TO ATLANTA

Mark your calendar for our 33rd Annual Conference, **August 3-6**, 2017, at the beautiful Marriott Buckhead in Atlanta, GA!



Co-hosted by Emory University and Sibley Heart Center, this year's conference will offer all of the features that people come for every year, including workshops, special programs for children, teens, and young adults; updates on the latest advances in research and treatment; free medical assessments (application required); and the opportunity to connect with others who are on the same medical journey.

If you attend, you can also participate in:

- Our National Walk for Victory to raise awareness and strengthen our community
- New workshops that focus on quality of life issues
- Enhanced networking opportunities which will enable you to increase your connections within the Marfan and related disorders community
- New and exciting activities and trips for teens, children, and young adults

OUR ANNUAL CONFERENCE OFFERS SOCIAL AND EDUCATIONAL PROGRAMS FOR ALL AGES, FROM FIELD TRIPS FOR CHILDREN TO RESEARCH UPDATES FROM THE WORLD'S LEADING EXPERTS ON MARFAN SYNDROME AND RELATED DISORDERS.

CHECK [MARFAN.ORG](http://MARFAN.ORG) FOR REGISTRATION INFORMATION IN THE SPRING

# SPRING GALAS—CELEBRATING COMMUNITY

THE **MARFAN** FOUNDATION

# HEARTWORKS

*St. Louis*

SATURDAY  
**MARCH 4 2017**  
FOUR SEASONS HOTEL  
ST. LOUIS

*Honoring*

**Christina Gurnett, MD, PhD** Associate Professor of Neurology, Washington University School of Medicine—2017 Distinguished Research Award ✦ **Dana Gima, RN** and **Barb Stehman, RN** nurse coordinators for the Marfan Syndrome and Related Disorders Clinic at Washington University and Barnes-Jewish Hospital—2017 Hero with a Heart Award ✦ **Cheryl Marshall** Marfan Syndrome and Related Disorders Clinic at Washington University and Barnes-Jewish Hospital—2017 Spirit of Service Award ✦

# HEARTWORKS

# *Gala*

WEDNESDAY  
**MAY 10 2017**  
GRAND HYATT NEW YORK

HONORING  
**Joseph Coselli, MD**  
Vice Chair  
Michael E. DeBakey  
Department of Surgery  
Baylor College of Medicine  
**Sinclair Li**  
Executive Vice President  
CBRE

4TH ANNUAL MARFAN VICTORY GALA ✦ SATURDAY, APRIL 22, 2017 ✦ MINNEAPOLIS AIRPORT MARRIOTT HOTEL  
Master of Ceremonies: Andy Erikson Featured speaker: Dr. Anjali Bhagra will address stress management and resiliency training

# WE REMEMBER AND HONOR

We are grateful to our members and friends who have made contributions in memory of, or in honor of, the following individuals. These donations are fully appreciated and support our programs and services that create a brighter future for all those living with Marfan syndrome and related connective tissue disorders.

## Donations In Memory of:

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 Elias Adame  
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 Dr. Michael Beardslee  
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## Donations In Honor of:

All Marfan Folks  
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 Steven Bankert  
 Dr. & Mrs. Charles A. Berst  
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## **Congratulations, New York City Marathoners!**

We are so proud of our runners in the 2016 New York City Marathon! They raised more than \$75,000 for the Foundation's life-saving programs and services! We are in awe of your spirit, determination, commitment, and positive attitude.

Our 2016 NYC marathoners were: Sara Maher and Bryan Butvick (pictured here with their daughter's name on their headbands), Johnsie and Rob Garrett, Hillary Sapanski, Ezra Jennings, Cara Brobst, Jon Gould, and Lisa Lore.

Thank you to our runners and all of their supporters. It means so much to our community to know that others care about them and are willing to take on a challenge on their behalf.

To learn about other ways to be a community fundraiser, please turn to page 12.