CONNECTIVE **ISSUES**

WINTER 2014

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.

Contents

CONNECTIVE **ISSUES**

WINTER 2014
VOLUME 33 | NUMBER 1

THE MARFAN FOUNDATION
22 MANHASSET AVENUE
PORT WASHINGTON, NY 11050
516-883-8712 | 800-8-MARFAN
WWW MARFAN.ORG

COVER: TEAM VICTORY MEMBER LISA JEFFRIES, OF COSTA MESA, CA, DURING THE LONG BEACH MARATHON ON OCTOBER 13, 2012. READ ABOUT TEAM VICTORY AND WALK FOR VICTORY ON PAGE 14.

NEW NAME, NEW LOOK, MOVE FOUNDATION FORWARD

Dear Friends,

Welcome to our new *Connective Issues* magazine, which shares stories that impact our community. Like our former newsletter, you will receive this magazine three times a year. In addition, in the Fall of 2013, we began to distribute a monthly newsletter by email, with news on timely happenings in our community.

This year has been quite remarkable. At our annual family conference in Los Angeles, we proudly announced that we are now The Marfan Foundation. Our new brand, which includes a refreshed logo and a new tagline, better reflects who we are and better positions us for accomplishing our mission to create a brighter future for people with Marfan syndrome and related disorders.

The Marfan Foundation is an evolution of our organization's previous name, the National Marfan Foundation. It places the focus on Marfan syndrome, and it is our goal to make "Marfan" a household word. Our new tagline, "Know the Signs. Fight for Victory." reflects our goals for early diagnosis and proper treatment so that people living with Marfan syndrome and related disorders can live a long and productive life. It also inspires people to take action and join us in the fight for victories over Marfan syndrome and related disorders.

Complementing our new name and tagline is our contemporary new logo, which is comprised of two interlocking links that represent our collaborations and connections with patients and families, the medical community, and corporate and government partners. The logo also symbolizes our work with Marfan syndrome and related disorders, which have overlapping features and overlapping treatments. You can read more about branding and nonprofits on page 9.

We will not rest until we've achieved victory—a world in which everyone with Marfan syndrome or a related disorder receives a proper diagnosis, gets the necessary treatment, and lives a long and full life.

I hope that you are as excited about this next step for the Foundation as I am.

Sincerely,

Carolyn

Carolyn Levering
President & CEO

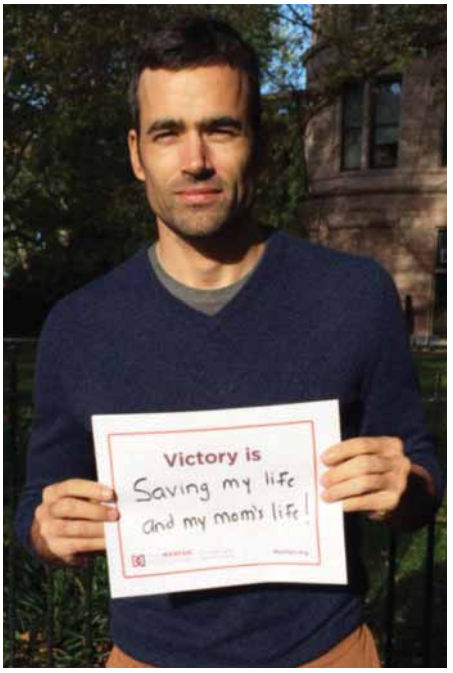
MARCH 1 HEARTWORKS ST. LOUIS **MARCH 22** HEARTWORKS WESTCHESTER **APRIL 10** HEARTWORKS GALA NYC

"Victory is to wake up every day, knowing that I am blessed to be alive, after an aortic dissection. Before that day, I had no idea what Marfan was. I know now. I am alive. That is victory for me."

– MAYRA KOHLER, FACEBOOK FAN

WHAT IS VICTORY?

Spending more time with family, finding a new Marfan friend, having hope, saving lives. These are a few of the ways people defined victory during the 2013 annual family conference when we revealed our new brand and tagline, "Know the signs. Fight for victory." What does victory mean to you?



MARFAN COMMUNITY PLAYS KEY ROLE IN RESEARCH BREAKTHROUGH

DR. LYNN SAKAI, CENTER, HAS DEVELOPED A NEW BLOOD TEST THAT MAY HELP EMERGENCY ROOM PHYSICIANS IDENTIFY DANGEROUS AORTIC ANEURYSMS AND DISSECTIONS.

It was nearly 10 years ago that our Professional Advisory Board member Lynn Sakai, PhD, a biochemist and researcher from Shriners Hospital for Children, set up an area at our annual family conference in St. Louis to collect blood samples from people with Marfan syndrome. In the Fall, the results were realized as Dr. Sakai and her colleagues published their paper about an innovative blood test that may provide a faster, simpler way for emergency room doctors and others to diagnose and monitor potentially deadly aortic aneurysms and aortic dissections (a tear in the wall of the aorta) for which early diagnosis is critical for survival.

The study, which was conducted with funding from The Marfan Foundation, was published in the prestigious *Circulation Research* journal. Shriners Hospital for Children and the National Heart, Lung, and Blood Institute also provided funding.

The research from Shriners Hospital for Children and Oregon Health & Science University, Portland, and Baylor College of Medicine and the Texas Heart Institute in Houston, found that high blood levels of fibrillin-1, a protein essential to the make-up of the body's connective tissue and blood vessels, are about twice as common in people with thoracic aortic aneurysm than in people with other types of aortic aneurysms. The high fibrillin-1 levels most likely are caused by damage to connective tissue or blood vessels. The researchers also found that high levels of fibrillin-1 fragments are more likely to be associated with aortic dissection, the life-threatening tear in the aorta. These new findings are potentially revolutionary because they mean that fibrillin-1 could someday be used in a blood test to diagnose aortic aneurysm and dissection. Currently the diagnosis is made through medical imaging, such as echocardiography, MRI, and CT scanning.

According to Dr. Sakai, whose research group first cloned the gene for fibrillin-1 (the Marfan gene), the original goal of this study was to provide better management of aortic disease for children and adults with Marfan syndrome, which puts people at up to 250 times increased risk of aortic dissection as compared to the general public.

"These findings are significant for public health because they represent the first human data to show that fibrillin-1 in blood could be a biomarker for thoracic aortic aneurysm and dissection," said Dr. Lynn Marshall, an epidemiologist from Oregon Health & Science University and the lead author of the study.

Marfan syndrome puts people at up to 250 times increased risk of aortic dissection

In addition, according to Dr. Susan Hayflick, Chair of Molecular and Medical Genetics, Oregon Health & Science University, "This is an excellent example of how research aimed at a rare genetic disorder helps people with similar diseases that are common in the general population. It is also an excellent example of how a multidisciplinary team of investigators can pool their expertise and resources for translational research that can directly enhance patient care."

"We are so grateful to our community, who not only help to provide financial support for innovative research studies, but also answer the call to action to participate in these studies," said Josephine Grima, PhD, Vice President of Research and Legislative Affairs for The Marfan Foundation. "Without their assistance, it would be impossible to a establish this new blood test."

MARFAN FOUNDATION AWARDS 2013 RESEARCH GRANTS

In the 1970s, the life expectancy for someone with Marfan syndrome was in the 40s. Less than thirty years later, due to early diagnosis, new medications, and advances in surgery, the life expectancy for people with Marfan syndrome was in the 70s, nearing the life expectancy of the general population.

In the past decade, research has accelerated even faster, giving our community great hope. What we have accomplished to date—with the collaboration of the research community and the support of the Marfan syndrome and related disorders community—is remarkable. We must continue our relentless pursuit of research so that life-threatening issues related to the heart and blood vessels can be eliminated and quality of life, which continues to be impacted by lung, eye, and skeletal problems, can be improved.

So many researchers are working hard to create a brighter future for people with Marfan syndrome and related disorders. Here are our latest grant recipients, selected after a rigorous review by our Scientific Advisory Board.

Our senior researchers

Lynn Sakai, PhD, Shriners Hospital for Children and Oregon Health & Science Center, is working to identify potential new drug targets by investigating new signaling pathways that work alongside the pathway known to contribute to Marfan syndrome.

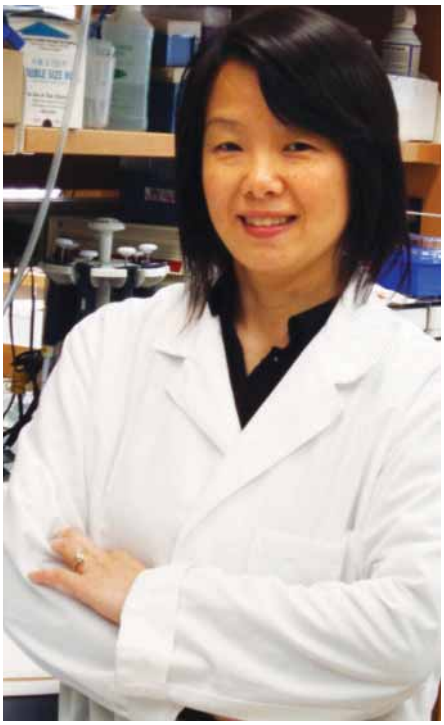
Chen Yan, PhD, University of Rochester, is looking at the role of smooth muscle cells in the degeneration of elastic fibers, which weakens the aortic wall and leads to aneurysm and dissection. This may lead to novel strategies for therapeutic intervention.

Rachel Kuchtey, MD, PhD, Vanderbilt Eye Institute, is working to characterize the development of glaucoma in a Marfan mouse model. She is also testing whether or not losartan, a promising treatment under clinical trial for Marfan syndrome, can either reverse or prevent the development of glaucoma.

Our early investigators

Venkateswaran Subramanian, PhD, University of Kentucky, is looking at the interaction between calpain and filamin. Increased calpain activity is highly correlated with filamin A degradation and aortic dilation in Marfan patients. This study is using a novel mouse model system to investigate whether or not this interaction could contribute to the degradation of the aortic root and cause aneurysms.

CONTINUED ON PAGE 6



CHEN YAN, PHD, IS LOOKING INTO THE ROLE OF SMOOTH MUSCLE CELLS AND THE WEAKENING OF THE AORTIC WALL.

GET INVOLVED IN **RESEARCH**

The participation of the Marfan syndrome and related disorders community in medical research is critical to advancing knowledge on the cause and treatments for these conditions.

Participating in a study may also provide you with more tangible benefits:

- You or your child may gain access to a treatment that is not available yet.
- You or your child may have a chance to see extra doctors or find out more facts about the medical condition.
- A study may connect you with other families going through the same medical challenges that you are experiencing.
- A study may offer closer monitoring or additional testing for you or your child, which may not be part of regular care.

To find out about the studies that are currently looking for people to enroll, visit **Marfan.org/get-involved**.

To me there has never been a higher source of earthly honor or distinction than that connected with advances in science.

– Isaac Newton

2012 RESEARCH UPDATES

Here are updates on last year's research grant recipients

George Tellidies, MD, PhD, Yale University
Dr. Tellidies is studying TGF-b signalling in vascular smooth muscle cells, which make up the aortic wall. Through genetic manipulations, he has inactivated TGFBR2 receptors in smooth muscle cells in Marfan mice. His findings suggest that basal TGF-b signaling in smooth muscle maintains postnatal wall homeostasis and impedes aortic disease progression.

John A. Elefteriades, MD, Yale University
The objective of this study is to develop a novel blood test for diagnosing and monitoring aneurysms and to predict impending aortic dissection or rupture in both non-syndromic and Marfan and related disorder patients. To date, patient recruitment and sample collection have begun.

Jay D. Humphrey, PhD, Yale University
This is the first engineering analysis of the physical stresses that occur in the aorta and result in aortic enlargement and eventual rupture. All indications thus far support the hypothesis that an overproduction of glycosaminoglycans (GAGs) could both disrupt normal wall maintenance and initiate aortic dissections. Additional experiments are planned to confirm these initial findings and, if confirmed, could lead to a new therapeutic target in Marfan syndrome.

Rajan Jain, MD, University of Pennsylvania
Dr. Jain's research is aimed at understanding the identity and embryonic origin of the cells composing the aortic valve and aorta. His research suggests that cells from a common embryonic origin, the cardiac neural crest, contribute to the formation and maturation of both the aortic valve and aorta. Insults to this vulnerable pool of cells during human development may lead to both problems in valve formation and aortic vessel maturation. Dr. Jain's research team is investigating the cues that regulate these cells and how these processes go awry to create the pathology commonly seen in patients with Marfan syndrome and related disorders.

2013 RESEARCH GRANTS continued

Hadas Shiran, MD, Stanford University, is trying to develop a new MRI method to improve the ability to detect thoracic aneurysms and non-dilated aortas that are at risk of rupturing. This would help patients and their medical teams pick the safest, most effective time for their surgeries.

Michael Fischbein, MD, PhD, Stanford University, is uncovering new insights into the mechanisms of ascending aorta enlargement in Marfan syndrome using microRNAs. This could help shift the current approach to aortic root aneurysm from a "diagnose and treat" approach to a "predict and prevent" method.

Our fellowship researchers

Sarah J. Parker, PhD, Johns Hopkins University, is investigating a possible miscommunication that occurs in different cell types that make up the aorta and cause it to enlarge. She will study whether or not correcting this miscommunication can help reduce the aneurysm in a mouse model of Marfan syndrome.

Josephine Galatioto, PhD, Mount Sinai School of Medicine, is working to prevent the life-threatening complications of vascular disease by identifying the druggable factors (known or predicted to interact with drugs) that may promote early aneurysm formation in Marfan syndrome.

DR. HADAS SHIRAN IS WORKING TO DEVELOP A NEW MRI METHOD THAT WOULD HELP DETERMINE THE BEST TIME FOR SURGERY.

MEDICAL QUESTIONS & ANSWERS

Dr. Richard Devereux, New York Presbyterian Hospital, a member of our Professional Advisory Board, answers questions provided by our members.

At what point do you decide if the aortic root is dilated or if there is an aneurysm?
Aortic dilatation (enlargement) is recognized when the diameter of a segment of the aorta falls above the upper end of the normal range, which generally puts about 2% of people without obvious disease above the upper limit of normal. Many physicians caring for people with Marfan syndrome grade dilatation as mild, moderate, or severe enough to warrant consideration of surgery. This graded approach is more suitable for long-term care of patients than an approach where someone abruptly changes from not having an aneurysm to having one at a specific aortic size. At present, available data suggests that replacing a slowly dilated aortic root in Marfan patients at a diameter of about 5 cm prevents enough aortic dissections to offset the risk and symptoms of heart surgery within a few years, and also provides a high likelihood of being able to have successful valve-sparing surgery.

Is a transesophageal echocardiogram (TEE) better than other imaging techniques for viewing the mitral valve?
TEE is better under very specific circumstances:

1. When there is reason for the cardiologist to think that mitral regurgitation may be more severe than detected by the standard technique of transthoracic echocardiography, TEE can be helpful because it can see parts of the mitral valve (mitral regurgitant jets) that might be hidden by the left lung during a standard echocardiogram.
2. When a patient is being considered for mitral valve repair, a TEE can help assess the likelihood of successful surgical repair or determine whether or not a patient may be a good candidate for innovative catheter-based techniques for valve repair without needing open heart surgery.

If you have successful elective valve-sparing aortic surgery, what is the likelihood that you will need additional aortic surgery in the future?
There are three components to the answer to this question:

1. With careful patient selection, a very small proportion of patients (about 1%) need surgery within the first months to a year.
2. After a replacement of the aorta closer to the heart, there is a very low risk (but not zero) of the need for additional surgery on the aorta further away from the heart. About 1.5 percent of patients need this additional surgery, regardless of whether or not they have the aortic valve replaced in the first surgery.
3. About 20 percent of patients have more than mild leaks across their aortic valve after valve-sparing procedures, which are generally stable during short-term follow-up. However, it is unknown if these patients and others may have later deterioration of their preserved valves, which have been placed in the unnatural environment of a supporting dacron graft.

Do tattoos interfere with the visualization of MRI and CT scans?
No.

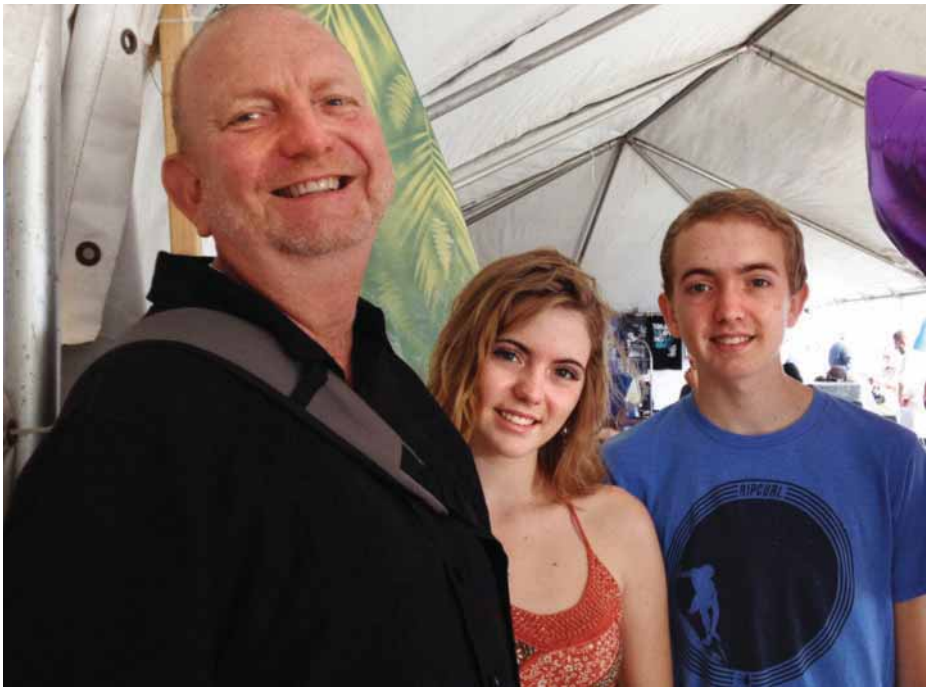
RESOURCES AND ANSWERS FOR YOU

If you have questions about various aspects of Marfan syndrome and related disorders, our website is your go-to place on the internet. Under "Resources & Answers," we have comprehensive information, including free downloads about Marfan syndrome and related disorders, as well as other tools for patient care. There is also a section of resources for schools. Many of these downloads are available in Spanish as well.

In addition to the print resources, our website features Q & A videos, in which medical experts on Marfan syndrome and related disorders answer the most common questions that people with Marfan syndrome and their families ask.

If you have further questions, use the "Ask a Question" form on our website or contact our help center at support@marfan.org or call 516-883-8712 x126. Amy Kaplan, the nurse in our help center, can provide personalized responses to your questions. Note that our regular business hours are 9:00am–5:00pm Eastern Time.

SURVEY FINDS ONE IN FIVE PATIENTS HAD SURGERY BEFORE GETTING A DIAGNOSIS



KEVIN SONGER, OF PALM COAST, FL, WAS DIAGNOSED WITH MARFAN SYNDROME AT THE AGE OF 54, WHEN HIS AORTA DISSECTED. HE PREVIOUSLY HAD MANY OTHER SURGERIES DUE TO WEAK CONNECTIVE TISSUE. AFTER HE WAS DIAGNOSED, HIS CHILDREN, JINCY AND RUAIRI (PICTURED HERE) WERE ALSO DIAGNOSED WITH MARFAN.

One in five people with Marfan syndrome had some form of surgery before they were diagnosed with the potentially life-threatening condition, according to a survey of 1,277 people we conducted from July 21–August 18, 2013. We conducted the survey to identify the signs that lead to a Marfan syndrome diagnosis and better understand the diagnosis process that people go through.

Alarmingly, of those who had surgery before they were diagnosed, 20 percent had an operation to repair a tear in their aorta, the large artery that takes blood away from the heart. In addition, 27 percent had an operation on tendons, ligaments, or joints; 24 percent on their back; 22 percent on bones; and 14 percent to repair a chest deformity.

> "It is concerning that so many people had surgery to repair their aorta before they got their Marfan diagnosis"

"It is concerning that so many people had surgery to repair their aorta before they got their Marfan diagnosis," said Alan C. Braverman, MD, Director of the Marfan Syndrome Clinic at Washington University School of Medicine and Chair of our Professional Advisory Board. "If they had been diagnosed with Marfan syndrome first, they could have undergone preventative aortic surgery before the aorta dissected. Many patients do not survive acute aortic dissection. People with Marfan syndrome who undergo surgery for an enlarged aorta may often expect to live a normal lifespan. However, the long-term outcome after an aortic tear occurs is not nearly as favorable."

Among people surveyed, the features that most often raised suspicion of the condition were skeletal abnormalities, especially long limbs (67%); long, flexible fingers (63%); flexible or extremely loose joints (51%); greater height than other family members (43%); chest bone that either sinks in or sticks out (42%); and curvature of the spine (33%).

Other features that raised a red flag were unexplained stretch marks (28%) and a dislocated lens in the eye (23%).

"The diagnosis of Marfan syndrome is based on a collection of characteristics, and many of them are common in the general population. Together, however, they could indicate an underlying problem that affects the heart and blood vessels and can be life-threatening," said Dr. Braverman.

Getting the diagnosis for Marfan syndrome is not always easy because it requires several tests done by different specialists: an echocardiogram or CT scan by a cardiologist, a slit-lamp eye exam by an ophthalmologist, and a skeletal exam by an orthopedist. Usually, a medical geneticist is also involved. While nearly half of people surveyed indicated that their diagnosis was confirmed in three months or less, almost one in five said it took them seven months or longer to receive a confirmed diagnosis.

We will highlight these findings in our medical education programs throughout the year.

TO HELP RAISE AWARENESS SO PEOPLE RECEIVE A TIMELY DIAGNOSIS AND PROPER CARE, VISIT Marfan.org/get-involved

VOLUNTEERS MAKE A DIFFERENCE

NEW TOOLKITS DEBUT IN 2014

February is Marfan Awareness month, the time for everyone in our community to do their part to help create a brighter future for people living with Marfan syndrome and related disorders. This year, it's even easier to raise awareness and raise money for education, patient support, and research with our new toolkits, which take you step-by-step through the process for planning and implementing a successful event.

ANDREA WITTE ORGANIZED AN EVENT AT HER SCHOOL TO RAISE AWARENESS AND GENERATE SUPPORT FOR THE FOUNDATION. YOU CAN TOO!

Andrea WItte, a high school student in Anchorage, AK, is one of our many dedicated volunteers who has held successful fundraising and awareness events. What she's found most successful is combining local awareness events at her school with an online personal fundraising page on FirstGiving. As part of her most recent school-based event, she raised more than $4,000 by sharing her story online and promoting her FirstGiving page when she promoted her event.

"It was important to me to have an event that raised both awareness of Marfan and raised money for the Foundation," said Andrea. "It feels great that people who attended the event shared the information with others who suspect they have Marfan and a few are currently getting evaluated. And the FirstGiving page gave my friends and family who couldn't be there a way to support my efforts."

You can hold a successful fundraising and awareness event, and there's no better time do it than in February, which is Marfan Awareness Month. The time to plan is now. The new toolkits on our website can help you with events in schools and doctors office, restaurants and businesses, and online. It there isn't one that meets your needs, you can contact us for personal assistance at volunteer@marfan.org.

Please join us and participate in Marfan Awareness Month by spreading the word and saving a life. Know the signs. Fight for victory.

TOOLKITS NOW AVAILABLE

AWARENESS TOOLKITS

Adopt-a-School Awareness
Awareness Table
Medical Office Awareness
In My Hands Documentary
Marfan Awareness Month Social Media

FUNDRAISING TOOLKITS

Fundraising on the Web
Dine for Dollars
Workplace Giving
Fun Friday School Fundraiser

You can download these toolkits from marfan.org. Just click on Get Involved > Volunteer > Raise Awareness and Fundraise. Questions? Contact volunteer@marfan.org.

NONPROFITS AND **BRANDING**

Most people associate "branding" with well-known companies like Nike, Coke, and Apple. So why do we, a nonprofit, need to have a "brand?"

Branding is about an organization's identity—its vision and mission, values, personality, and general communications. We at The Marfan Foundation understand that to achieve our organizational goals and help our community, it is critical that we present our organization in a way that has the greatest impact. According to research by Stanford University on the role of the brand in the nonprofit sector, "a strong brand is increasingly seen as critical in helping to build operational capacity, galvanize support, and maintain focus on the social mission."

Our brand was developed based on research, what you our constituents shared with us in surveys, one-on-one interviews, and other communications. We listened to your feedback and incorporated it into the strategies that are propelling us forward in everything that we do—in research, in support, in education and awareness.

Through a unified voice, a focused mission, and the strength of our community, we continue our relentless pursuit of a brighter future for everyone living with Marfan syndrome and related disorders.

SPECIAL THANKS TO

The team at Cedars-Sinai Medical Center that spearheaded coordination of the conference: Dr. Robert Siegel, cardiologist, Dr. Ora Gordon, medical geneticist, Mitchel Pariani, genetic counselor, and Tami Kendra. (Pictured above: Mr. Pariani, Dr. Siegel, and Dr. Gordon).

Our Professional Advisory Board, chaired by Alan Braverman, MD, Washington University in St. Louis, for their participation in our conference clinic, medical presentations, and workshops.

Our Los Angeles community group of volunteers, led by Roelina Berst, Regine Wood Bluestein, and Suzanne Bowman.

Our Spanish language expert, Dr. Juan Bowen, Mayo Clinic, Rochester, MN.

Our volunteer photographers, Santino Hunter and Frank Papini.

Our corporate supporters:
Cedars-Sinai Medical Center
Medtronic Endovascular
St. Jude Medical
Terumo Cardiovascular Systems
Univision Communications, Inc.

FRANK PAPINI, ONE OF OUR VOLUNTEER PHOTOGRAPHERS

ANNUAL FAMILY CONFERENCE

TRIED AND TRUE, OLD AND NEW, COMBINE TO CREATE VALUABLE EXPERIENCES FOR NEARLY 500 ATTENDEES

JOSIE VILLARRUBIA OF CHICAGO IS HELPING THE MARFAN FOUNDATION REACH OUT TO SPANISH-SPEAKERS, A NEW PROGRAM THAT WE PILOTED AT OUR CONFERENCE.

The Marfan Foundation's 2013 Annual Family Conference, co-sponsored by Cedars-Sinai Medical Center, was a great success, with nearly 500 members of our Marfan syndrome and related disorders community coming together for a weekend of information and support, laughter and tears, advocacy and empowerment. Participants made new friends and reconnected with old friends. Children, teens, 20-somethings, and young adults bonded with their peers in ways that were both educational and fun. We offered special workshops for older adults, unaffected spouses, and parents, ensuring that everyone had an opportunity to have their questions answered.

For the first time, we held a Spanish-language program for Hispanic members of our community, giving many families an opportunity to learn in their native language. In collaboration with Univision, we secured extensive media coverage about Marfan syndrome and related disorders in Spanish-language media to raise awareness even beyond the conference. We are happy to make the conference and our materials accessible to more people in our community so that they too can be empowered to advocate for themselves and get appropriate care.

HIGHLIGHTS OF CONFERENCE MEDICAL PRESENTATIONS

Duke Cameron, MD, Johns Hopkins, addressed aortic surgery in children.

Surgery is recommended for children when their aortic diameter is greater than 5.5 cm. If they have a family history of aortic rupture or dissection, the threshold (when surgery is recommended) is lowered to 5.0 cm. In addition, if they have rapid enlargement (greater than 1 cm per year) or progressive aortic insufficiency (abnormal function of the aortic valve that results in the leaking of blood from the aorta back into the left ventricle of the heart) with moderate enlargement, then surgery is recommended.

Surgery is also necessary if a child has an acute or chronic dissection; however, this is very rare in children under 12 years of age. In contrast, mitral regurgitation (the back flow of blood from the left ventricle to the left atrium of the heart through an abnormal mitral valve) is more common in children than adults. If mitral valve repair and root replacement surgery is necessary in a child, adult-size devices are used so it is rare that a child "outgrows" an operation.

If your child needs aortic surgery, Dr. Cameron notes that:

- Prophylactic aortic root replacement (surgery before there is a tear or rupture) is a safe operation that can prevent aortic rupture and dissection.
- Long-term results with composite grafts are excellent.
- Valve-sparing operations have excellent results for the short-term (long-term follow-up results are not yet available for this relatively newer procedure).
- Late distal aortic dissection (tears in the aorta further away from the heart) and arrhythmias remain challenges to long-term survival.

Reed E. Pyeritz, MD, PhD, addressed genetic testing in Marfan syndrome.

The most recent diagnostic criteria for Marfan syndrome give a greater weight to genetic testing (FBN1 testing) in the diagnostic assessment than before.

Many laboratories in North America (and elsewhere) perform genetic testing and insurance often covers part or all of the cost. It usually takes two to three weeks to get the results.

For a person who meets the clinical criteria, finding a mutation in FBN1 is not necessary. However, some people and many physicians take comfort in having the clinical diagnosis confirmed by the genetic test. For a person who has some but not enough clinical features, especially a young person, genetic testing can be useful. It is also useful for the parents of a diagnosed individual to have genetic testing. However, it is important to know that not finding a mutation does *not* exclude the diagnosis. That's because new mutations for Marfan syndrome are relatively common (25–30%).

For a person who has some but not enough clinical features, especially a young person, genetic testing can be useful.

Genetic testing can also be useful for a couple planning to become pregnant. Prenatal diagnosis and pre-implantation genetic diagnosis coupled with in vitro fertilization are possible.

When it comes to genetic testing, remember:

- Acquiring your family medical history, which is simple and does not require any expense, should be your first step.
- Molecular testing can save considerable costs of pre-symptomatic clinical screening.
- Molecular genetic testing is not always simple. People should have pre- and post-test counseling.

David Liang, MD, PhD, Stanford University Center for Marfan Syndrome, discussed important points to be aware of after you've had aortic valve-sparing surgery.

- The valve still needs to be watched to make sure it continues to function properly.
- Valve replacement is needed when the pattern of growth changes or when the valve begins to leak.

After you have had an aortic dissection repaired:

- Life expectancy is good.
- Continued medical treatment is needed.
- Close monitoring is needed (continued vigilance!).
- Appropriate intervention may be needed (potentially additional surgery on other parts of the aorta).

If you have a dissection of your descending aorta, surgery is not necessarily needed. Instead, doctors use aggressive blood pressure control to stabilize the aorta. This involves getting the blood pressure below 110 mmHg and then monitoring the aorta regularly (3, 6, 9 months) until the dissection is stable. Then, life-long vigilance is needed.

AND THE AWARD GOES TO...

WE GRATEFULLY RECOGNIZE THE CONTRIBUTIONS OF THE FOLLOWING GROUPS AND INDIVIDUALS TO CREATING A BRIGHTER FUTURE FOR PEOPLE WITH MARFAN SYNDROME AND RELATED DISORDERS

Antoine Marfan Award
Dr. David Rimoin (posthumous)

Cheryll Gasner Spirit of Service Award
Eileen M. Ilberman, Old Saybrook, CT

The Priscilla Ciccariello Award
Roelina & Chuck Berst, Los Angeles, CA

Heart of the Matter Award
Rene Jones, Merrimack, NH

Rising Star Award
Andrea Witte, Anchorage, AK

TOP: CHUCK AND ROELINA BERST ACCEPTED THE PRISCILLA CICCARIELLO AWARD FROM CAROLYN LEVERING.
BOTTOM: CAROLYN LEVERING PRESENTED THE HEART OF THE MATTER AWARD TO RENE JONES

Volunteer Recognition Awards
Alpha Phi Omega—Theta Upsilon Chapter, Case Western University, OH
Hannah Blackwell, Schaumberg, IL
Stephanie Begley, Boston, MA
Laura Biggart, DeWitt, IA
Campbell Family, Oilton, OK
Justin Clark, San Francisco, CA
Nakia Cole, Wills Point, TX
Teri Dean and Stella Lane, Pleasant Hill, IA
Rachel Epperson, Fort Worth, TX
Frank Garcia and Matthew Garcia, Los Angeles, CA
Beth and Jon Gould, Fox Point, WI
Peter Grabel, Greenwich, CT
Scott Griebel, Dover MA
Amber Harbison, Trussville, AL
Hannah Michelle Harris, Shelby, NC
Santino Hunter, Los Angeles, CA
Janet Jenkins, Grayson, KY
Heidi Law, San Rafael, CA
Jennifer Lynch, Mountain Lakes, NJ
Kathy Magee, San Antonio, TX
Tara McGuire, Castle Rock, CO
Alix McLean Jennings, Madison, NJ
Dominga Noe, Carmichael, CA
Frank Papini, Los Angeles, CA
Sara Paul, Staten Island, NY
Chris Sears, Medford, NY
Glenn Stidham, Nesconsett, NY
Molly and Matt Wowk, Verona, WI

Volunteer Recognition Awards: Teens and Kids
Peter Donato, Framingham, MA
Saffra Parks, Tulsa, OK
St. Paul's School, Concord, NH

Volunteer Recognition Awards: Group Leadership
Los Angeles Network Group:
Roelina Berst
Suzanne Bowman
Regine Wood Bluestein

Volunteer Recognition Awards: Groups
Birmingham Network Group
Dallas-Fort Worth Network Group
Detroit Network Group
Heart of Iowa Chapter
Massachusetts Chapter
New Hampshire-Vermont Network Group
Mid-Atlantic Chapter
Northern Illinois Chapter
Phoenix Network Group
Staten Island Network Group

Volunteer Recognition Awards: Tall Clubs
Portland Skyliners Tall Club
Paramount Tall Club of Chicago
Tall Clubs International Convenion

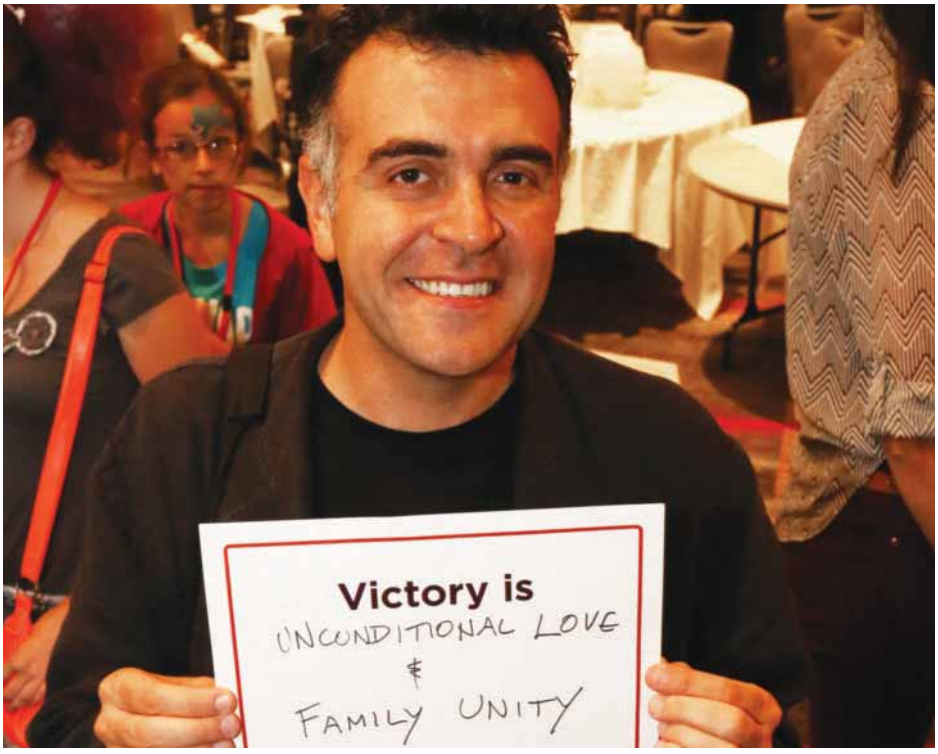
CAROLYN LEVERING AND DR. REED PYERITZ WITH ANNE AND ANNIE RIMOIN, WIFE AND DAUGHER OF THE LATE DR. DAVID RIMOIN, WHO ACCEPTED THE POSTHUMOUS AWARD.

LIVING SUCCESSFULLY

A highlight of our annual conference each year is the closing panel that features several members of our Marfan syndrome and related disorders community speaking about how they live successfully. Here are some of the highlights.

We are grateful that there is a place for our family to come to during those dark hours and moments of despair... I haven't met any group who was so genuine and compassionate towards each other until I met you. This group has a big heart, a great soul, and an amazing gift to lift each other up during those difficult days.

– SUELY JOHNS, VALENCIA, CA



DIRECTOR AND SCREENWRITER ERIC NAZARIAN MODERATED THIS YEAR'S LIVING SUCCESSFULLY PANEL

Marfan has taught me many things, among them that my passion and vocation was to be a social worker not a marine biologist. It's taught me not to take things for granted and to always have a plan b, c, and d. It's taught me that one person can make a difference and a group as strong as we are can save many lives.

– JOSIE VILLARRUBIA, CHICAGO, IL

As a child, I wanted to become a fighter. Who would have known then that a fighter is exactly what I would have to become. I fought for my life in hospital beds when my lungs were collapsing. Twice. I've fought the grief of losing close family members, including the father that taught me so much. I'm fighting a battle right now against an aorta that is threatening to tear. But you know something, for all these battles, I know I've got the strongest support that anyone could ask for: my family, my friends, all of you here today.

– JOSH LIM, ORLANDO, FL

WALK FOR VICTORY

WALK FOR VICTORY IS A FUN WAY FOR PEOPLE OF ALL AGES AND ATHLETIC ABILITIES TO GET INVOLVED, RAISE AWARENESS, AND GENERATE SUPPORT FOR THE FOUNDATION.

Our new program, Walk for Victory, focuses on community and puts the FUN in fundraising! In the Spring of 2014, we will have walks in Phoenix, Dallas, Long Island, and Boston. Have you been looking for a fun and easy way to volunteer for The Marfan Foundation? Looking for a way to contribute that can involve all members of your family, young and old? Then, walking for victory is for you.

Each Walk for Victory will include a short walk, and by "short," we mean less than a mile. It is a noncompetitive event that focuses on community and everyone wins by participating. You can even participate in a wheelchair or pushing a stroller. After each walk, there will be music and games, as well as refreshments for all who participate.

Once you register for the walk of your choice, you will be prompted to set up your own personal fundraising webpage. This will enable you to capture your story about how you, or someone you know, is affected by Marfan syndrome or a related disorder and share it with your friends, family, and coworkers. By explaining why this cause is so important to you, you can raise awareness and vital funds to support the Foundation's life-saving programs and services.

There is no cost to participate in this event, but we do ask that each walker brings in at least one donation of any amount. You can make a donation yourself or you can collect donations on your personal fundraising webpage. For example, if your family of five wants to participate in a Walk for Victory, set up your personal fundraising webpage and get at least five people to make a donation.

By registering for the walk, you receive many helpful and fun benefits:

- Your own personal or team fundraising webpage
- A bi-weekly email which includes fundraising tips, event updates, inspirational stories, and incentives for the top fundraisers
- Snacks, games, entertainment, and prizes at the event
- The opportunity to connect with others in your community
- Satisfaction in knowing that you are helping to create a brighter future for the thousands of people living with Marfan syndrome and related disorders

Bonus: Every walker who raises at least $100 will receive an exclusive Walk for Victory t-shirt.

For more information, please contact victory@marfan.org.

Would you rather run?

Join Team Victory, the Foundation's endurance program and participate in the Rock n' Roll San Francisco Half Marathon on April 6, 2014. Participants will run 13.1 miles through beautiful San Francisco to the beat of bands and entertainment throughout the course. Register for this race at: firstgiving.com/marfan/2014SanFran

Check out the other races in the 2014 Race Schedule at Marfan.org/get-involved.

JOIN US AT A WALK FOR VICTORY NEAR YOU AND HELP US RAISE AWARENESS AND ADVANCE OUR LIFE-SAVING WORK:

Phoenix Walk for Victory—Sunday, March 23, 2014
Dallas Walk for Victory—Saturday, April 26, 2014
Long Island Walk for Victory—Saturday, May 3, 2014
Boston Walk for Victory—Saturday, May 10, 2014

To learn more and register, visit Marfan.org/get-involved

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:

Adrian Adame
Carol Adame
Elias Adame
Tommy F. Arthur
Johnny Bardin
Carol Abbatiello Bartnick
Zoe Mae Bayouth
Arthur N. Berven
Anniell R. Bevis
Madison Blaire Boudreaux
Sharon Brake
Paulie Burke
William Callahan
Bill Campbell
Benjamin Mark Carlson
Sarah Cayo
Thomas & Anna Ciccariello
Galen K. Clark
Francis Iredell Clarke
Mary Clayton
Kimberly Ann Cobb
Christopher Copeland
Liam Faegen Corcoran
Clifton Durand
Julie Jennifer Emmons
Joseph E. Fabiszewski, Sr.
Christopher Scott Faust
Marcia Bunde Filip
Sean Flaherty
Gabby
Joseph R. Gagliano, Jr.
Charles Gilmore
Christopher Gilmore
Gregory Gilmore
Todd Glidden
Catherine Goodzey-Roper
Joseph G. Gorman, Jr.
Kerry Greenwald
Mark Grimes
Mildred Hansen
John Heinemann
Shawn Heldt
Nathan Blake Hensley
Frank Hergenreter, Jr.
Barbara Hildenstein
Jim Hinds
RuthAnn Holp
Patricia Hurley-Burger
Barbara Harden Johnson
Eileen M. Ilberman
Julie Kurnitz
Shirley Langer
Jeannine Lee
Wendy Linder
Ricardo L. Lowry, Jr.
Alexandrea Macaluso
Mabel Marie Maines
Glinda "Paulette" Belcher Marshall
Leona Martin
John Mascaro, Sr.
Claire R. Michaud
Frank W. Miller, Jr.
Flora R. Kaplan Mincer, MD
Patricia Moran
Sandy Janeen Morris
Peter C. Mosshart
Paula Richards Neil
Barbara Ann Newton
Michael Andrew Norton
Vincent "Jerry" O'Loughlin
Dr. Michael Palmer
Stephen Parfenoff
Linda Diane Parman
Christopher Paulsen
Espirito Perez
Carol Pijoan
Jackie Prindle
Renee Lynn Randant
David A. Ricca, M.D.
M. Brock Robertson
Frank Romano
Maryann Roney
Adele Schell
Gene Schweitzer
Spencer Sellas
Joseph Sinay
Steven
Bridget Stewart
Les Swiggum
Matthew A. Tish
Curt Van Ess
Jeff Walbridge
Barbara J. Wayman
Wendy Weiss
Eric L. Wika
David M. Willson
Jeffrey James Wurst

Donations In Honor of:

Charles & Roelina Berst
Austyn Bevis
Edith Black
Jerry Bluestein
Patrick Bowman
Tommy Cadden
Louise Chudnofsky
Finnegan
Henry Floyd
Marty Goldberg
Mildred Goldstein
Heidi Green-Stepp & Shayna Green
Aaron Hartmann & Liz Keenan
Maggie & Roy Hattersley
Lynn Holleran
Ross & Kathleen Hooker
Cassie Jennings
Daniel Keyes
Gordon Keyes
Benjamin Kuehn
Jerry Lerman
Ann Lowenfels & Steve Redler
Meaghan & Thomas Lynch
Paul Lynch
Kyle Mann
Lois B. Meyer
Dr. Craig Miller
Karen Murray
Ryan O'Sullivan
Delaney Pasteka
Brooke Pulliam
Lisa Roberts & Ryan Larson
Peter Roos
Mr. & Mrs. Stan Ross
Joey Tish

DR. JESSICA DAVIS HONORED

DR. JESSICA DAVIS WITH SOME OF OUR YOUNG MEMBERS AT ONE OF OUR RECENT CONFERENCES.

Congratulations to Dr. Jessica Davis, a long-time member of our Professional Advisory Board, for receiving the Excellence in Human Genetics Education Award from the American Society of Human Genetics (ASHG).

The award recognizes an individual for contributions of exceptional quality and great importance to human genetics internationally. Awardees have had long-standing involvement in genetics education, contributions in more than one area, and contributions of substantive influence on individuals and/or organizations.

Dr. Davis, who is Associate Professor of Clinical Pediatrics at Weill Cornell Medical College and Associate Attending Pediatrician at both New York-Presbyterian Hospital and the Hospital for Special Surgery in New York City, is a past recipient of The Marfan Foundation's Antoine Marfan Award for her many contributions to our community.

The Marfan syndrome and related disorders community is fortunate to have such a dedicated and caring medical geneticist as a clinician, advocate, and friend.

THE MARFAN FOUNDATION

22 MANHASSET AVENUE
PORT WASHINGTON, NY 11050

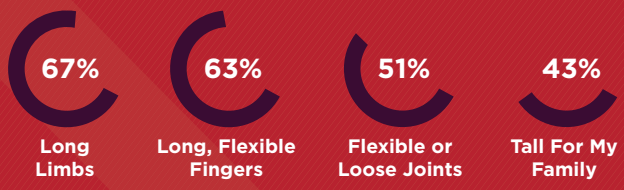
Read more about the survey findings on page 8

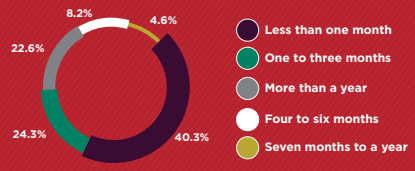
We hope you like our new *Connective Issues* magazine! We want to make sure you receive the magazine, and our monthly newsletters, by email. To make sure you don't miss our news, please send your email address to us at publicity@marfan.org.

THE MARFAN FOUNDATION
Know the signs. Fight for victory.

Did you know that skeletal features are key in raising suspicion of Marfan syndrome?

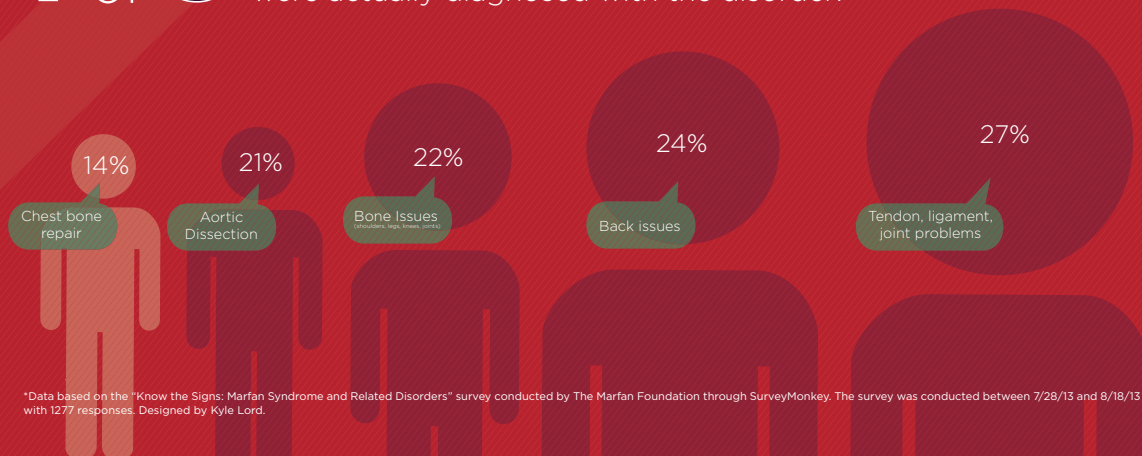
There are many outward signs that should raise the suspicion of Marfan syndrome. The most common are features in the bones and joints.





40% of people got their diagnosis in less than a month, but for nearly 25% it took more than a year.

1 out of 5 Nearly 20% of patients said they, or a family member, had operations that were likely due to Marfan syndrome before they were actually diagnosed with the disorder!



*Data based on the "Know the Signs: Marfan Syndrome and Related Disorders" survey conducted by The Marfan Foundation through SurveyMonkey. The survey was conducted between 7/28/13 and 8/18/13 with 1277 responses. Designed by Kyle Lord.