

# matters of heart

issue3volume3 fall2011

A publication for people affected by Loey's-Dietz syndrome, but not defined by it.

## boardnotes



Joseph Galli, Director and Co-Founder of the LDSF-Canada and Gretchen Oswald, President of the LDSF, raise awareness about LDS at the 2011 Canadian Marfan Association annual conference in Halifax.

As the end of the year approaches, we look forward to what the rest of 2011 will bring and eagerly await a new start in 2012. Entering this season of family and thankfulness we want to take a minute to be thankful for the things that matter most...

our LDSF friends and family who continually believe in and support our mission and work tirelessly to grow our organization. We are thankful for you... each of you... for being a part of our community in big and small ways.

As we look forward, we want to call upon our community to support the LDSF as 2011 comes to a close and to consider how to give support — both time and resources — in the upcoming year.

### yearendgiving

It takes money to do what we do... to connect families impacted by LDS with one another, to work to spread awareness amongst the medical community about LDS and to seek out new ways to impact research and treatment plans for our families.

We are thankful for every donation and fundraiser this year. And now we're asking you to consider supporting our mission and making a year-end donation. All contributions are tax deductible. Will you consider supporting us this holiday season with a gift of \$10, \$25, \$100 or more to help those impacted by LDS? Donations can be made online at [www.loeydietz.org](http://www.loeydietz.org) or by mailing a check made out to the LDSF to: P.O. Box 22468, Baltimore, MD 21203.

We thank you in advance for taking heart and supporting the LDSF with your finances.

### boardofdirectors

The LDSF Board of Directors is a group of volunteers who dedicate their time, talents and efforts to help guide, support and direct the mission of the LDSF. The Board has the responsibility of ensuring that our organization is moving closer to our stated goals, setting direction for future projects and overseeing the finances of our non-profit.

The LDSF is looking to grow our Board of Directors in 2012. Board Members will serve two-year terms, be required to participate in quarterly teleconference meetings and one in-person meeting per year, and to serve on various subcommittees as assigned and appropriate. Board members do not have to have or be related to someone with LDS, but must have a passion for helping encourage education, foster research and provide support for those impacted by LDS.

Currently our Board of Directors is in need of individuals with skill sets and passions in the following disciplines: Public Relations/Media, Fundraising/Grant Writing, Law, Medicine, Nonprofit and IT.

If you or someone you know is interested in learning more about becoming a member of our Board of Directors, contact Gretchen Oswald ([Gretchen.oswald@loeydietz.org](mailto:Gretchen.oswald@loeydietz.org) or 443-850-5874) for more information or to request an application packet.

Again, as we reflect on all that we have to be thankful for in 2011, we are thankful for each of our donors, volunteers and members. Without you, we would not be able to do what we do. From the bottom of our hearts we say THANKS!

*Until next time,  
The LDSF Board of Directors*

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## foundationmission:

The Loey's-Dietz Syndrome Foundation (LDSF) is a 501(c)(3) non-profit organization dedicated to:

- encouraging education about LDS and related connective tissue disorders for medical professionals and lay communities in order to aid in identification, diagnosis and treatment of Loey's-Dietz syndrome
- fostering research about LDS
- providing a support network for parents and families affected by Loey's-Dietz syndrome

## mattersofheartmission:

To provide an outlet for members of the LDSF community to connect and stay informed with accurate, relevant and timely information



# heartrenderings

*Victor Kovac and his three children have LDS. Since his initial emergency surgery, Victor has had seven major and several minor surgeries and hospitalizations related to complications of LDS. His hope is that sharing some of his family's experiences may be of help to others dealing with LDS. This is the first part of his story.*

Late August 1993 found me standing in a crowded market street outside of the Port of Mogadishu, Somalia, with a weapon in my hand as I supervised crowd control at the entrance to the UN-controlled port. This was a few weeks prior to the now famous "Blackhawk Down" incident. The conflict between the Somalia militia forces of the warlord Mohamed Farrah Aidid was heating up between the Somalia and UN force with sniper fire, mortar attacks, grenade attacks and remote detonated mines occurring with increasing frequency. I remember being extremely alert trying to take in my surroundings, peering intensely into the darkened windows of the nearby buildings and watching the faces of the people in the streets — just another day in my eight-year, active-duty Army career.

Suddenly I felt an intense pain between my shoulder blades as if someone had stabbed me in the back with a knife and my knees buckled. I had no idea what was happening to me but I knew that if I fell down in the middle of that street that I could very well be kidnapped. I was leading a small group of Somalia Port Police and the squad on the gate were Arab infantry from the UAE. I could barely communicate with them and I did not trust them to protect me if I became incapacitated. The only other American near the gate was inside the wire working. I felt very light-headed but, after the initial stabbing pain, things got better and it became more of a deep throbbing pain. Not wanting to leave the young American mechanic alone, I waited another ten minutes while he finished his work and then headed back into the port complex. I was wracking my brain trying to figure out what was happening to me — was it some kind of sciatic nerve problem or a slipped disc? I had stupidly lifted my footlocker, personal gear and rucksack all at once a few days before when I took the helicopter taxi to the port and maybe it was that...or maybe the Arab cigarette that I bummed from one of the guards on the gate had poisoned me...it tasted like formaldehyde.

I was in a bit of dilemma as I had an important city-wide security meeting that day at UN HQ and it was critical that I be there. My transport with a Pakistani police squad was already arranged and waiting, so I hopped in their vehicle and rode to the airport where I was to catch a helicopter taxi to the UN compound on the outskirts of the city.

It only took about five minutes to arrive at the helipad. As I waited I became increasingly agitated as the pain was growing in intensity and now had radiated through my chest and was becoming increasingly impossible to ignore.

In retrospect I must not have been thinking clearly as instead of seeking aid from the English speaking New Zealanders who ran the heliport I decided to walk the kilometer or so along the bluffs overlooking the Indian Ocean to the Pakistani compound to get a ride back to my unit. As I trudged along, the pain continued to increase, so I started handing my gear to my soon overloaded bodyguard — first my AK-47, then my heavy flak vest, then my

helmet — keeping only some intelligence documents that were classified non-foreign. After a bit of difficulty communicating with the Pakistani gate guard I caught a ride back to the port and checked in at our aid station.

I am not sure how much time had elapsed from the initial tear at this point but I imagine it was around an hour. The pain had become almost intolerable. I remember my vision had narrowed down to a tunnel of sorts with no peripheral vision at all. I told the young medic Sergeant that if he didn't want to see a Captain start crying I needed something for the pain and soon.

A German Army chiropractor happened to be passing through the port that day picking up some of their vehicles. He took a look at me and had me lay on a table thinking that he could "adjust" the back pain away. I was willing to try anything that offered a chance at relief from the pain that was making it harder and harder for me to think. Fortunately the US medic was very sure and very firm in refusing help from the German officer, and he said "I am sorry Sir but I can't allow you to treat the Captain. I know you are a doctor in the German Army, but the US Army doesn't recognize chiropractors, and the Captain's blood pressure is all wrong. The high is too high and the low is too low."

The Sergeant had me Medevaced to the US Army 46th Combat Support Hospital at the University of Mogadishu compound. I don't know that young Sergeant's name but I owe my life to his calm and intelligent assessment and fast action.

The field hospital was housed in a number of tents and was limited in available diagnostic equipment and experience with this type of trauma. An ultrasound was inconclusive, and a chest x-ray, EKG and arterial blood gasses were all normal. The extreme pain, blood pressure and a III/VI systolic and diastolic murmurs were not normal. The surgeons at the field hospital made a diagnosis of "aortic regurgitation caused by a dissecting aortic aneurysm" and started treatment to lower my systolic pressure while arranging for air evacuation through Germany to Walter Reed Army Medical Center in Washington, DC. It took over 24 hours before I could depart Somalia by a US Air Force air ambulance.

The last thing I remember as the plane took off was looking out the slowly closing ramp at the beautiful, strikingly savage African coast where the desert meets the Indian Ocean. I lost consciousness before the ramp closed all the way and really remember very little of the twelve hour flight to Germany.

It is a long way from the Horn of Africa to Washington, DC. By the time I reached the refuel point in Germany it was some 36 hours after the initial pain. The air evacuation team and the young Army doctor who accompanied me from the field hospital decided to move me from the airfield to Landstuhl Army Medical Center. Further evaluation there showed that I had a 7cm aortic root aneurysm. Due to a continued delay in the departure of the air evacuation transport from Germany and fear that I would not survive the seven hour flight to Washington, it was decided to seek medical assistance from a German surgeon since the United States didn't have any surgeons at Landstuhl that could or would undertake the surgery needed to repair the aneurysm.

*continued next page*

# heartrenderings continued

Fortunately for me one of the Army doctors at Landstuhl knew a German civilian surgeon at The University of Kaiserslautern Hospital who had studied similar procedures at one of the big US medical centers in Houston. There on August 22, 1993, I underwent my first big surgery, termed a Bentall procedure, during which my aortic valve was replaced with a #27 Bjork-Shiley Valve and aortic graft. A post-operation TEE showed dissection of the descending aorta that would need repair at a later date. Although the German doctor conferred with me prior to the operation, I must have had some anesthesia-related memory loss because I don't remember anything that happened from my departure from Somalia until I awoke after the operation in the German hospital ward.

Once stabilized, I was transferred back to Landstuhl Army Medical Center and remained there a few days awaiting further evacuation to Walter Reed. After a week or so of bed rest at Walter Reed the Army began the process of discharging me and sent me back to my duty station at Ft. Eustis, Virginia. Despite the German surgeon's warning that further surgical repair of the descending aorta was needed, the Army told me that everything was repaired and I shouldn't have any additional problems. Just a few months later I found myself discharged on the Temporary Disability Retired List (TDRL) with no job and with a greatly reduced pay level. Worse was the fact that what I considered my calling, my career as an Army officer that had started twelve years earlier, was gone and forever beyond my reach.

Psychologically it was a dark time. My wife had been eight months pregnant with my youngest son and on bed rest from difficulties related to the pregnancy. We were jobless and far away from our families. We

were also troubled that I had no idea what had caused the aneurysm. The Army doctor who attended my evacuation from Somalia later wrote a paper based on his experience with my case that was published in the journal of Military Medicine, "Acute Dissecting Aortic Aneurysm in an Operational Environment." In this paper he wrote "Subsequent evaluation has revealed no apparent cause for his propensity to develop aneurysms, including Marfan syndrome, syphilis, rheumatoid spondylitis and atherosclerosis."

This fundamental lack of understanding for what was happening to me and the resultant problems with knowing how to treat what was going on — or even to know if anything ongoing was happening — would create a problem both in my personal treatment and the growing but completely unknown and unsuspected health problems faced by my three children, who were all pre-school age at the time of my initial surgery. This darkness of knowledge would last for some twelve years until LDS was identified and described by Doctors Loeys and Dietz.



The Kovac family — Victor, Jacob, Jesse, and Adair — is proud of the way they work together to deal with the issues that arise from all having LDS.

*We're sharing the Kovacs' story in several parts. Be sure to watch your inbox in the coming weeks for our email blasts sharing more of this family's journey with LDS.*

## congratulations

Hopkins Children's Center gastroenterologist Anthony Guerrero, M.D., Ph.D., has received the George Ferry Young Investigator Development award of the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition (NASPGHAN) Foundation for his research on childhood digestive diseases. The award, which comes with \$150,000 in research funding over two years, is given to outstanding scientists who study the origins, mechanisms and treatments of a wide variety of pediatric gastrointestinal disorders and is based on his proposal entitled "The Role of Transforming Growth Factor Beta in Eosinophilic Gastrointestinal Disorders and Inflammatory Bowel Disease."



Dr. Guerrero's clinical practice focuses on eosinophilic intestinal disease (esophagitis, gastroenteritis, and colitis) as well as inflammatory bowel disease especially in the context of immunodeficiency. He also has a special interest in intestinal problems of those affected by Loeys-Dietz syndrome. He was honored at the NASPGHAN/NASPGHAN Foundation Awards Reception Friday, October 21, 2011, during the annual meeting in Orlando.

Congratulations Dr. G., and thank you for all you continue to do for our LDS families!

## raisefunds

LDSF heart pins by artist Terri Kerchner continue to be available and make great gifts for Mother's day, teacher gifts or any occasion! Pins are \$15, plus shipping and handling. Please email [Kate.Jurgens@loeysdietz.org](mailto:Kate.Jurgens@loeysdietz.org) for more information.

We now offer cut-out LDSF TakeHeart hearts that can be used in multiple ways to help generate heartfelt monies. Hearts can be used for a "hang-a-heart" type campaign where for a specific donation amount an individual can write their name on a heart and have it displayed on the wall at a school, store, church, or wherever! Create your own campaign to help us raise money and awareness for the LDSF. Please email [info@loeysdietz.org](mailto:info@loeysdietz.org) to request hearts for your own campaign.

*The LDSF is a registered non-profit corporation and is exempt from the United States Federal Income Taxes under section 501(c)(3) of the Internal Revenue System. All gifts are tax-deductible. We thank you in advance for your kind assistance and support.*



LDSF pins by Terri Kirchner





LDSF kids pose with the Baltimore Orioles mascot at the 2010 LDSF conference.

# save the date

## LDSF Conference 2012

June 29 - July 1, 2012 Baltimore, Maryland

Save the date for Friday, June 29 through Sunday, July 1 to gather in Baltimore, Maryland, with other LDSF families. Participate in large and small group sessions, get up-to-date medical information and spend time with others impacted by LDS. More details coming soon.

# quentin's quest 3 update



Hello all! My web site has been updated and now includes a lot more information on my current quest and the quests of years gone by. Please visit my site at [quentinsquest.org](http://quentinsquest.org) and check out the new photos as well as a guestbook that you can sign!

I hope all of you are collecting your spare change. There is less than two months left before Thanksgiving and I can't wait to see how we did. The Loeyes-Dietz Syndrome Foundation is working on some cool stuff to help inform doctors and parents about the condition that makes me so unique. They can use every penny, so please remember to toss your coins into your bins every night before you go to bed, take your change to the bank and send me a check made out to the LDSF.

Thanks for all your help!  
Quentin

# european friends

Three families impacted by LDS and living in various places in Europe took advantage of a holiday and met up in Algarve, Portugal. The group spent a short time together encouraging one another and sharing their LDS experiences. "It was quite emotional for all of us as this was the first time meeting other LDS families," said Rachel Martin, the group organizer. "We had a good time together but it was too short. We hope we can meet up again soon in the UK or Ireland!"

The group is interested in connecting with others in Europe. To connect with this group, email Rachel at [Rachel.Martin@loeysdietz.org](mailto:Rachel.Martin@loeysdietz.org).



L to R: Rachel (LDS), Elly (mom to Beatrice), Basil (dad to Rachel), Milena (mom to Ricardo), Daphne (mom to Rachel).  
In Front: Beatrice and Ricardo

# last chance

Dr. Paul Sponseller and his team at the Johns Hopkins University are interested in obtaining more information on bone health in individuals with Loeyes-Dietz syndrome. Entitled "The risk of osteoporosis and skeletal fractures in patients with Loeyes-Dietz Syndrome (LDS)," the questionnaire aims to look at history of fractures (broken bones), soft bones and other orthopedic concerns.

This is your last chance to participate in the study. If you have not yet done so, check out [www.loeysdietz.org/research.php](http://www.loeysdietz.org/research.php) for more information on how to participate.

# LIVE new orleans update

On Wednesday, August 24, more than 150 people turned out at the 12 Bar on Fulton for the first annual LIVE! New Orleans fundraising event. The night raised more than \$10,000 for the LDSF to help encourage education, foster research and provide support for those impacted by LDS.

"Family, friends and the community all pulled together to raise funds and awareness for LDS. It was a night to remember!" said LDSF member Anne Accardo, who hosted the evening.

"The planning committee was amazing and worked so hard on every detail," she said. "It was a classic, southern reception. Everyone filled the room, mingled and enjoyed the amazing food and wine that was donated by local favorite restaurants."

The celebration continued with a welcome and LDSF information presented by LDSF Board member Kate Jurgens and a live performance by the Brass-A-Holics band.

"It was a night I know I will never forget," said Accardo. "I'm already looking forward to next year!"



## McKesson Canada Client Advisory Committee

San Francisco, CA; August 24-25

In the United States, McKesson operates RelayHealth which provides connectivity solutions that help consumers and healthcare organizations securely exchange information. McKesson Canada is implementing a similar system in Canada where all of a patient's pertinent medical history, test results and prescriptions will be stored in one place online. Currently a large technology team is customizing this collaborative platform for the Canadian market.

McKesson has formed a Customer Advisory Committee (CAB) to seek direction and input for the Canadian market and asked the LDSF-Canada to participate. LDSF-Canada is the only advocacy group represented amongst the provincial heads of health across the country. LDSF-Canada and McKesson are working on a pilot program whereby LDSF-Canada will be the first advocacy group to offer the system to its members, planned for implementation in 2012.

## Maritime Connective Tissue Clinic CME Day

Halifax, Nova Scotia, Canada; September 21

The Continuing Medical Education (CME) day at the Maritime Connective Tissue Clinic attracted 120 people, where the LDSF-Canada and the LDSF had a booth. Seventy-five medical kits and LDS "head-to-toe" checklists were distributed. The LDSF spoke on a panel that discussed the importance of genetic testing, representing the patient's perspective.

Members of LDSF-Canada had dinner with Dr. Gabrielle Horne, director of the clinic, and two of her colleagues to discuss sharing best practices in the treatment of the LDS population. Dr. Horne operates a multidisciplinary connective tissue clinic that sees approximately 400 patients per year in the Maritime provinces.

## Canadian Marfan Association Annual Conference

Halifax, Nova Scotia, Canada; September 22-23

Approximately 30 people attended the annual Canadian Marfan Association's conference where Gretchen Oswald, genetic counselor and President of the LDSF, spoke on LDS and led a discussion group on LDS with Dr. Timothy Bradley. The LDSF and LDSF-Canada had a booth and distributed 20 medical kits to attendees.



## International Congress on Human Genetics

Montreal, Quebec, Canada; October 12-15

The International Congress on Human Genetics, hosted by the American Society of Human Genetics (ASHG), attracted more than 6,500 genetics professionals from all over the world. The LDSF-Canada was selected by the Genetic Alliance as one of 10 advocacy groups to participate in their Advocacy Track, a program focused on raising awareness in this community. High level meetings with industry officials took place with leaders of the LDSF-Canada, and more than 30 medical kits and DVD's were distributed in one-on-one meetings.

At the conference a number of testing labs gave approval to include a letter directing patients who have been diagnosed with LDS to the LDSF website.

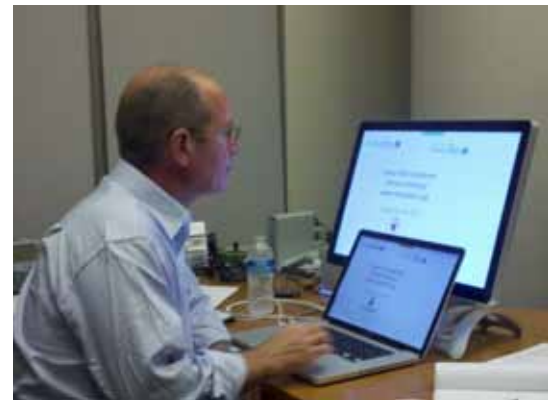
## Genetic Counsellors of Canada Annual Conference

Montreal, Quebec, Canada; October 16

More than 120 participants from across Canada came together for this annual conference, where the LDSF and LDSF-Canada gave out 68 kits and DVD's and where we were invited to speak with students completing the Genetic Counsellors program in Canada.

On September 29, Joseph Galli (LDSF-Canada) and Gretchen Oswald (LDSF) organized a webinar for the Shriner's Hospital Network (both the USA and Canada) to present about LDS and raise awareness. Dr. Hal Dietz and Dr. Paul Sponseller (head of orthopedics at The Johns Hopkins University) made a 90-minute presentation to the Chiefs of Staff for the Shriners Network. The first 60 minutes was a presentation by the doctors, followed by a 30-minute question and answer session.

Dr. Peter Armstrong, Chief Medical Officer of the Shriners Network of Hospitals allowed the LDSF to use his monthly management meeting to communicate the importance of LDS to his medical staff. The Shriner's Network sees thousands of orthopedic cases per year and, as a result of the webinar, can now better identify patients with LDS.



## thankyou

Thanks to everyone who participated in our conference survey and volunteered of their time and resources to help the LDSF. We are thankful to work with such a wonderful group of individuals who believe in our mission and support our efforts.

It's not too late to volunteer! If you're interested in helping out the LDSF, email [volunteer@loeysdietz.org](mailto:volunteer@loeysdietz.org) and get involved.



Joseph Galli, Director and Co-founder of the LDSF-Canada, represented the LDSF-Canada and the LDSF at various conferences during the fall raising awareness about LDS in the medical community.

# doctorsdinner

Francoyse and Sal Fratino, directors and co-founders of the LDSF-Canada, hosted a private dinner at their home with Dr. Hal Dietz and Dr. Bart Loeys on October 14 in Montreal.

The Fratino's, and the LDSF-Canada, hosted 50 people, including participants from Japan, Australia, the United States and Canada. Five LDS patients attended the evening along with medical professionals from across the specialties.

The evening was intended to raise awareness about LDS and on-going research efforts. Each guest received an LDSF pin, a medical kit and a copy of the Shriner's webinar presentation by Dr. Dietz and Dr. Sponseller (see "Shriner's Webinar," previous page). As a result of the evening we now have offers from many additional medical centers to help aid both Foundations.



# awarenessjapan

Joseph Galli, director and co-founder of the LDSF-Canada, met with the Morisakis from Japan to discuss promotion of LDS in their country. The Morisakis manage the National Cardiovascular Center and Research Institute in Osaka and have over 70 confirmed LDS patients in their care.



## upcoming events

### Forum on Rare and Orphan Diseases

Montreal, QC, Canada; November 26

LDSF will participate in a panel discussion in Montreal for the Quebec Rare Disease network. The objective of the day is to discuss solutions to help overcome the obstacles faced by those who are affected by a rare disease and their families in our health system. Joseph Galli will give a testimonial entitled, "Taking Matters in Your Own Hands."

### Crossing the Finish Line for Caroline

Bossier City, LA; March 3

The second annual Crossing the Finish Line for Caroline walk/run event will take place on Saturday, March 3, 2012, in Bossier City, Louisiana. More information will be available soon at [loeysdietz.org/events.php](http://loeysdietz.org/events.php).

*Interested in planning an event to help raise funds for the LDSF? Have an idea? Contact us at [info@loeysdietz.org](mailto:info@loeysdietz.org) and let us help you start an event or check out the Event Request form on our web site ([www.loeysdietz.org/events](http://www.loeysdietz.org/events)) to get started.*

## contributions

The Loeys-Dietz Syndrome Foundation is able to continue to Take Heart thanks to the generous support of individual donors. We welcome all levels of donation, whether it's a one time gift, a monthly donation, or the establishment of a matching gifts program. The Loeys-Dietz Syndrome Foundation is grateful to its members and friends who have made generous contributions in memory of or in honor of their loved ones. These donations are fully appreciated and support our mission. All those who gave generously will be acknowledged at the end of the year.

