

matters of heart

issue3volume2 fall2009

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



Dr. Dietz spends time with LDSF kids during a summer mini-conference in Baltimore, MD.

founders notes

As this newsletter is written, the foliage is reaching its peak of color, and with it comes the harvest of pumpkins, squash, and corn. Fall is here!! Seasons change and time brings about changes. Like the seasons, so too has our foundation seen wonderful growth and change. We are entering our third year, and we feel both enthusiasm for what we have achieved in '08, and '09, as well as the pressure to meet the expectations we set for 2010.

As we reflect back and look forward, we have a number of reasons to give thanks to our Board, members, and friends this fall!!

looking back

The first year our Board stepped up to the challenge in helping to define goals. Like now, they had lots of energy, and many fantastic ideas. Many families shared similar stories, and that inspiration offset the fact that the foundation was running on little funds. Despite our low-budget start-up, a website was put in place, which immediately put the organization into the homes of whomever could find us. The live forum and information on the site was our first real buy-in for members to see what our organization could offer. Although we had an identity, it became evident that we needed a new logo and branding. Enter year two, and the wonderful assistance of designer Megan Rhee!

Although we had 501 c 3 status, a mission statement, goals, and even a website, we were young! (We still are young!!) Still, families were anxious to support, and consequently, we had an

obligation to show members that its work was starting to produce results. We needed to keep the energy and enthusiasm levels high. Members and friends began to identify activities for which to fundraise, and this was made possible by the website and quarterly newsletter, and of course other platforms such as FaceBook. We also introduced online giving on our website, as well as through JustGive. We collaborated with the National Marfan Foundation to create a series of five fact sheets specifically for Loey's-Dietz, available not only on our website, but theirs as well.

With the continued growth, families began to have various fundraisers in several different states, memorials and honorariums were being made, and donations continued to trickle in. Most recently, we had an informal gathering in June in which families (and Board members) who had been working together could finally meet face-to-face, as well as hear some of the latest information on Loey's-Dietz at Johns Hopkins, catch an Orioles game, and of course take a Pirate ship ride in the Inner Harbor with new and old friends. There was a much larger response than what we had anticipated, and we were excited to see the enthusiasm to support an event that was a highlight of the summer!

looking ahead

We have made great strides in the last year. As we become more focused, have a committed Board, and an ever-increasing awareness of LDS, our various planned events, outreach and education initiatives will help continue our mission. This includes a mini-conference in Baltimore this summer. Families that attended in June expressed an interest

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foundation mission:

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 to 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

matters of heart mission:

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

Loey's-Dietz 
Syndrome Foundation

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“I’m recently diagnosed as LDS. I searched information on LDS through internet to find that little information is available in Japanese except for some academic papers; the information was mainly written as Marfan syndrome variants. I wanted information focusing on LDS using daily language everyone can understand.

Fortunately, I can handle English and could find LDSF, but many of LDS affected Japanese are not. When I found LDSF, I was so relieved; I found a place to belong!”

- **Yukako Ogura, Japan** Thanks to our presence on the web, LDSF is having a global impact! Yukako is working to translate LDSF materials into Japanese and desires to set up a chapter of LDSF in Japan to provide information and community in her home country. Welcome Yukako!



heartrenderings

Up until 3 years ago, our family, myself (46 yrs old), my oldest son, Ryan (21 yrs old) and my daughter, Chelsi (17 yrs old) were diagnosed as having Marfan Syndrome. Ryan was the only one with some of the Marfan characteristics - tall, long limbs, long fingers and toes. We all are double jointed, have pectus carinatum, enlarged aortas, easy scarring and bruising, just to name a few. It was very difficult dealing with doctors when we told them that we had Marfan syndrome. We were not the classic Marfan patient.

In February 2006, due to my increasing aorta, I underwent a Tyrone David Valve Sparing surgery at Stanford Medical Center. My aorta measured 5.2 cm and with “Marfan Syndrome” it was determined that it was time for the surgery. After the surgery, my cardiologist, Dr. David Liang suggested that we take a blood sample and send it to University of Washington to see if I had Loeys Dietz Syndrome instead of Marfan syndrome. I did have some characteristics of LDS including a bifid uvula, wide set eyes, cervical-spine instability and the aortic aneurysm.

About 8 weeks later, my diagnosis was confirmed. I was officially diagnosed with having Loeys Dietz Syndrome. Of course, I had very mixed feelings on the whole diagnosis, but it was a relief to know exactly what I had. During this time, I was still on the mend from surgery and also started my search for information on Loeys Dietz Syndrome. I felt like a sponge just absorbing every bit of information I could get my hands on. I knew that if I was diagnosed with Loeys Dietz, then the likelihood of my children having it was pretty high. They had some of the same characteristics as I did.

So, my children’s yearly check ups continued, just like we have done since birth with each of them. In 2007, it was discovered that my son’s aorta had grown to 4.5. Since my diagnosis was confirmed and they had suspected that my children also had LDS, it was time for Ryan to undergo his Tyrone David Valve Sparing surgery also. He was 18 years old and had a completely different outlook on the whole surgery. He knew what had to be done, so he just kept the mind set of, “Let’s go and get it done.” This did make the whole process a little easier since I didn’t have to spend time trying to convince him that everything was going to be ok. He had seen me go through this surgery and recover just fine. So, once again, I made a call to Stanford and told them, “Same surgery, different patient”. The surgery was scheduled for February 2008. Ryan’s surgery went as planned and he is now fully recovered.

Chelsi, my dear lovely Chelsi, was the next candidate for surgery. She was a senior at Monache High School in Porterville, CA and very active

with yearbook, photography, prom, football games and any other extra curricular activities she could find. Although she knew deep down inside that she had to get the surgery, it was not discussed much throughout the year. She went through a denial phase. The “I am not having it done phase”

and the “Why me phase?” I knew this was going to be a challenge. Since it was her senior year, I knew that I was not going to plan her surgery during the year, so the plan was right after graduation. So, around February of 2009, I started my calls to Stanford. Once again “Hi, this is Stacy Spain, I would like to set up the same surgery but different patient”. By this time, I was so relaxed with the staff at Stanford and they all made me feel so at ease and were eager to help in any way possible. This is a great help when scheduling such a traumatic event for you or a family member. The date was set for June 17, 2009. As time got closer to the event, so did prom, graduation, senior pictures, Disneyland trip and all the bells and whistles that come with being a senior in high school. She graduated on June 5, 2009. What a happy day that was. After we came back down to reality, we realized we only had about a week to get everything in order before we made our trip to Stanford Medical Center in Palo Alto, CA. Chelsi finally accepted the fact that she was going to have surgery and that she did. Her surgery went well and within 6 days she was home and ready to start her road to recovery. Through this process she and her brother have become so close now, they are both starting college in the fall together.

This is just a brief summary of our road to recovery and dealing with Loeys Dietz Syndrome. Three major surgeries in 3 years is a lot to handle, although I feel my family has learned a lot about what is really important in life. We cherish the time we have together. We have all become closer and more knowledgeable about LDS. To anyone that is facing surgery or trying to deal with the diagnosis, please know that you are not alone. Please reach out to someone to help you get through it all.

My thanks go out to all the staff at Stanford Medical Center including Dr. Craig Miller (surgeon), Dr. David Liang (cardiologist), Sunny Pellone, Julie Morris, and Judi Lachenmyer. And last but not least, I could have never got through this entire time in my life if it wasn’t for my husband, Robert Spain, my other son Austin, my father Bob Smith, my uncle Larry Wheaton (LDS) and all my close friends and family. My mother, Margie Smith and brother, Rocky Smith were never diagnosed with LDS as they passed away before LDS was discovered. These 2 absolutely amazing people are the reason I am as strong as I am today. They are my true angels.



The Spains - Ryan, Chelsi & Stacy

NMFconference

The National Marfan Foundation's Annual Conference was co-hosted by the Mayo clinic in Rochester, MN on August 6-9. Many families and loved ones of those with Marfan syndrome, Loeys-Dietz syndrome and other related connective tissue disorders came together to learn about their disorders, current medical practice, and exciting research! Thank you to Dr. Hal Dietz and Dr. Duke Cameron, cardiovascular surgeon, who led informational sessions on LDS! It was a great weekend of connecting with other LDS families! Thank you to the NMF for their continued support and collaboration for those impacted by LDS.

The NMF Annual Conference held at Mayo was the best experience our family could have ever asked for. This being our first year, we didn't know what to expect. Within the first 2 hours of the general session on Saturday, we had already decided to make this an annual event. We received so much information about LDS we were overwhelmed. The information alone on how to work with the school was amazing. This year we followed the formats supplied and sent them in. Within 4 days of school starting, we received a revised 504 Plan from the school. The information just made everything go so much smoother. We have also started a medical file with the emergency information from another break out session, something we never thought of doing.

We asked our nine year old daughter, Caslin, what she thought was the best part of the conference. "Making new friends" was her response. That was priceless. She was so happy to see all the other kids



Caslin Kasten & Gretchen Oswald

that looked just like her. No one even mentioned her long fingers.

We would like to thank all of the speakers, with a special thank you going out to Dr. Hal Dietz for all of his research into LDS. The research with the mice is invaluable! I can't tell you how much that conference helped me as a person to better understand what is going on. My husband always had a better grasp than me. Now I feel a little more confident with what is going on inside our daughter.

It would be great if the LDSF could also have an annual conference. We know, with no doubt in our minds, we would attend.

- **Mary Kasten**, Elk Mound Wisconsin



Gretchen Oswald & The Cali family of Venezuela

teencorner

What advice do you give to other LDS teens facing surgery?

My name is Chelsi Spain and I recently had the Tyrone David Valve Sparing Surgery at Stanford Medical Center in Palo Alto, California. I had the best doctor (Dr. David Liang) and surgeon (Dr. Craig Miller) anyone could ask for. How do I know this? I figure that if this surgeon can perform the same surgery on my mother in 2006 and my brother in 2008, and I see they are healthy now, had little to no complications, and were out of the hospital in no time, I am in really good hands.

I underwent the surgery on June 17, 2009 and was home on June 23, 2009. Granted, I was in some pain and was pretty exhausted but that is all normal. I continued to do my breathing exercises and take my pain medication as directed. Before I knew it, I was starting to be my old self real quick. I still cannot do a lot of things i.e., lifting, pushing, pulling, etc. but I am back to a normal routine now through out the day.

Any of you teens that are facing this surgery or multiple surgeries, my advice is to not panic! I know it is easier said than done. No surgery is exciting but remember, life is. Of course I worried about it all, but if I thought about it for too long, then I would stress myself out and no doctor wants you to be stressed before surgery. That will increase your blood pressure. Remember to keep a smile on your face and remember what great family and friends you have and do things to have fun before your surgery to keep your mind occupied. You will get through this and you will be just as beautiful or handsome as you have always been! You are beautiful on the inside and out. Keep your head held high and live life to the fullest.

If you would like to ask me any further questions about my surgery, my symptoms or just need some help getting through it all, please email me at: pink_piggies_05@hotmail.com. - **Chelsi**, 17 years-old

FUNdraising

Thanks to David and Sam Coren! In lieu of birthday gifts this Spring, they asked their friends to donate to the LDSF! Over \$500 was given in honor of the Sam and David. Thanks boys or (mad-scientists). Your generous spirit is awesome!



David & Sam Coren

awareness

On September 5, 2009, the second annual Hardie Family Pig Roast took place just west of Brantford Ontario. Approximately 250 people attended the event which aimed at raising awareness and education about Loeys-Dietz Syndrome.

Jeremy Hardie died unexpectedly in 2005 at the age of 32 leaving behind a wife and 2 young children. With the help of the Genetics and Cardiology departments at the Sick Kids Foundation it was determined that Jeremy had Loeys-Dietz syndrome. Many other family members have since been diagnosed including his brother, Jason and his son, Evan.



Dr. Tim Bradley, Gloria Hardie (LDS) and Nancy Slater, RN. Dr. Bradley and Nancy Slater work in Cardiology and have an interest in LDS, MFS and other connective tissue disorders

foundersnotescont.

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to have a more formalized event, with the same family feel of an action packed, well informed two-day event.

We are anxious for all of your help! We would like to implement a membership drive, and strengthen our database. We would also like to seek out grant funding for awareness and education initiatives.

Other plans for 2010 include the further development of educational fact sheets to add to the series, as well as the development of a book that may help in the preparation of cardiac surgery for parents and children, and the continued financial support of studies in the areas of pregnancy and GI/allergy issues in LDS. We will also continue to collaborate with service organizations, such as the National Marfan Foundation, organizing the LDS sessions for their annual conference in Houston July 8th-11th. We hope to see ya'll there, too!!

While there are challenges and changes impacting all of us, we thank you for your continued support. We look forward to the upcoming events and hope you can attend and benefit. We also hope you consider FUNdraising in an effort to help support the LDSF. For ideas or more information, please contact at info@loeydietz.org. **org.** As the holidays approach, you may consider sending a gift to the foundation in honor of or in memory of a loved one. You may also gift online by visiting our website loeydietz.org and clicking on the JustGive link, or by going to justgive.org and typing in Loey-Dietz syndrome foundation as your charity!

Sincerely, *Beth Utz and Gretchen Oswald*

researchnews



David Loch, PhD

David Loch is a Postdoctoral Fellow in the Institute of Genetic Medicine at the Johns Hopkins University of Medicine. He is developing mouse models of Loey-Dietz syndrome in order to test new treatment strategies.

To better our understanding of Loey-Dietz Syndrome we have recently created two mutant mouse models of this genetic disease. These mice harbor mutations in the genes encoding transforming growth factor-beta receptors 1 or 2 (TGFBRI/Alk5 or TGFBRII, respectively). Initial investigation of both mouse strains has confirmed recapitulation of the LDS vascular phenotype, with widespread arterial disease including arterial tortuosity, aneurysm and dissection. In addition, mutant mice demonstrate musculoskeletal abnormalities. Work by Drs Tony and Pamela Guerrero also confirms gastrointestinal pathology similar to that seen in LDS patients. Current experiments revolve around elucidation of critical disease processes to better our targeting of potential treatments. On this basis, we are also initiating pharmacologic trials in these mouse models of LDS with drugs that have proved successful in Marfan syndrome, including Losartan.



orderform

Help raise funds for education, research, and support initiatives for the Loey-Dietz Syndrome Foundation. Consider cards for holiday gifts or Christmas cards. Give a unique LDS heart pin as a gift or wear to your holiday party! It's also a great way to involve friends and families to join in supporting the LDSF!

CARDS: packs of 10 available for \$10.00. **HEART PINS:** \$15.00; each pin is unique and created for the LDSF.

Quentin Waid began a quest two years ago to help raise funds for LDSF. You can see the whole story on **quentinsquest.org**. With much help from friends all over the world, his cookbook with hundreds of delicious recipes is ready to go! Funds go directly to help support LDSF initiatives. Thanks to Quentin and the Waid family for all of their hard work. Get cooking and give one for a holiday gift!

COOKBOOKS: \$15.00.

Name: _____

Address for Shipment: _____

Phone/Email address: _____

# of Holiday Cards:	_____	(x \$10.00 per pack) = \$ _____
# of 2009 LDSF Design Cards:	_____	(x \$10.00 per pack) = \$ _____
# of LDSF pins:	_____	(x \$15.00 per pin) = \$ _____
		SUBTOTAL = \$ _____

Please add \$5.00 of shipping costs - per \$50.00 order: CARD/PIN SHIPPING = \$ _____

# of Quentin & Friends Cookbooks:	_____	(x \$15.00 per book) = \$ _____
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Please add \$6.00 of shipping costs per book: (___ books x 6) BOOK SHIPPING = \$ _____

GRAND TOTAL = \$ _____

Portions of each item are tax-deductible and receipt will be sent with order. Thank you for your support of the LDSF.

Please send order form and check/cash to: **Gretchen Oswald, 3805 Monterey Road, Baltimore MD 21218**

(Gretchen.Oswald@loeydietz.org) Please make checks payable to: LDSF.



Holiday Cards



2009 LDSF design



Assorted Heart Pins



Quentin & Friends Cookbooks