

matters of heart

issue2volume1

A publication for people affected by Loeys-Dietz syndrome, but not defined by it.



top: LDS families enjoying a night out in Boston.

bottom: Quentin and Mo spending time with Dr. Dietz

conferenceupdate

LDSF Families Gather in Boston

More than 40 individuals attended the LDS Panel with Dr. Dietz and Dr. Loeys at the 2008

National Marfan Foundation Conference in Boston, Massachusetts in July. They reviewed information about C-spine instability, vascular imaging, and surgical and medication management.

Dr. Dietz announced the creation of three mouse models of Loeys-Dietz syndrome that mimicked the findings in the human population. This is exciting news as it allows for therapeutic drug trials to be performed on mouse models and is an important step in LDS research.

Dr. Anthony Guerrero presented preliminary information surrounding the research he and Dr. Pamela Frischmeyer-Guerrero have been doing examining allergy and gastrointestinal problems in individuals with LDS. Thus far, they determined an increased finding of sinus and ear infections due to craniofacial abnormalities and not due to underlying immune deficiency. This led to a higher incidence of asthma due to the sinus disease. About 30% of survey respondents had food allergies (compared to a 6% finding in the general population). A subset had eosinophilic esophagitis (increased white blood cells in the lining of the esophagus) as a result of food allergies. Symptoms included abdominal pain, poor weight gain, vomiting, decreased appetite and esophageal spasms. When the food allergy is identified and eliminated, individuals tend to see improvement in

symptoms. Additionally, vitamin-D deficiency is being investigated. Results of the full study will hopefully be released in the next year.

Dr. James Black, vascular surgeon, also answered questions from both LDS and Marfan families regarding aneurysm management outside of the aortic root. Upon review of MRA information from individuals with LDS at Johns Hopkins Hospital, elongation of arteries was observed over time. Longer term follow-up is necessary to determine if this impacts aneurysm formation. There is no other "common" pattern of aneurysm development outside the aortic root for LDS individuals. Brain aneurysms have been rarely seen. For arteries in the abdomen (including superior mesenteric artery, iliac, renal and celiac arteries), general management summaries included potential surgery when and if the artery is twice the diameter of the normal sized, neighboring portion of the artery.

As well, on the living successfully with connective tissue disorders panel, Danielle Stapleton, a 28 year old medical student shared her story of getting the diagnosis of LDS earlier this year. This was after episodes of multiple pneumothoraces and a subsequent echocardiogram showing an aortic root of 5.3 cm. Her advice to "living successfully" included knowing your body and trusting the pains that are different, utilizing resources and support by your connective tissue families, assembling a good health team, and treasuring and appreciating friends and families who get you through the hard times with love!

Finally, congratulations to Dr. Bart Loeys, MD, PhD, who was recognized with the National Marfan Foundation's Antoine Marfan Award.

inthisissue:

- Conference Update
- Texas Fun
- In Loving Memory
- Quentin's Quest
- Teen Corner
- FUNdraising
- Baltimore Cook-in
- Ask the Expert
- Contributions
- Get Involved
- Raising Awareness
- Holiday Giving

foundationmission:

The Loeys-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 Loeys-Dietz syndrome
- fostering research about LDS
- providing a support network, for parents and families affected by Loeys-Dietz syndrome

mattersofheartmission:

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

Loeys·Dietz
Syndrome Foundation

“I’ve been through the many phases of any parent whose child has a medical condition: despair over the unexpected, frightened by the unknown, mourning over the loss of ‘perfection’ to gradual acceptance and, finally, the bliss of enjoying the mundane in life despite it all.”

- Raquel, LDSF Board Member



texasfun

Paying tribute to their mother, the late Wendy Kent Churchill, is not something new for the Kent-Churchill family, nor the town of Fort Worth, Texas; however, this year, family, alongside Kent Motor Company, the Fort Worth Cats, and many wonderful sponsors, hosted the annual crawfish boil and golf tournament. These two events in one weekend



Kendall, Jylian and John

directly benefited the Loeyes-Dietz Syndrome Foundation. They were the first two events to kick-off fundraising for the LDSF! The events were planned by John and Kendall Bilbow, Tabitha Forsythe, Corrie Churchill and her brother Will.

The Crawfish Boil was held at the Fort Worth Cats Stadium on Saturday, April 12th, 2008. Poo Live Crew entertained and encouraged the crowds to come and stay. They were an awesome band and people were begging for more, and asking about next year! The crawfish and vegetables were great too, and people were enjoying the endless pit of food and drink!!

Throughout the night there was a silent auction with prizes such as signed guitars, unique artwork, spa gift certificates, spa treatment for your vehicle gift certificates, sporting event tickets, etc. In addition, there were live auctions for roundtrip airfare to anywhere and other amazing prizes! Some of the LDSF heart pins were sold for a donation of \$15.00 each. The golf tournament followed on Sunday, April 13th. There was a silent auction, and other fun activities such as 5 card draw, longest drive, etc. The two events were very memorable, and we hope that the Wildcats invite our foundation back for a wonderful fundraising time! We are pleased to say that approximately \$10,000 was raised. Way to go Texas!! Thank you for showing us that like the Lone Star state, we CAN help to raise funds!!



Poo Live Crew entertains the crowd.

inlovingmemory:

Robert “Rob” Franklin Hogan

June 16, 1981 - September 8, 2008
Tax Accountant for Profiler Consulting. Loving husband of: Marla Gardner Hogan; loving son of: Michael & Denise Hogan

“Rob loved life and all those around him. He was very wise and mature beyond his years. He accepted his medical challenges with a special grace and dignity. Rob was a remarkable son and loving husband. He was greatly appreciated and deeply loved by his brothers and sisters. He was a kind and cheerful friend.”

Special thanks to Dr. Joseph Coselli and staff of St. Luke’s Houston, Texas for several miracles.



Kayleigh Lorraine Palmer

November 15, 1989 -
September 27, 2008
United Kingdom

Much loved daughter of Louise & Andrew
Dearly missed sister of Craig
Devoted stepdaughter to Andy
Loved by all family and friends
In our hearts forever



Our fondest thoughts and well wishes go out to the families and friends of the Hogans and Palmers.

quentin’squest

Quentin’s Quest Part 1 has come to an end. Quentin raised more than \$12,000 from more than 450 individuals by asking for everyone he knew to mail him a dollar to support Loeyes-Dietz syndrome research.

Funds were split among the Loeyes-Dietz Syndrome Foundation, Make-A-Wish and All Children’s Hospital. Quentin presented a check to Dr. Hal Dietz at Johns Hopkins on May 13th.

Quentin’s Quest Part 2 is now underway as Quentin puts his love of cooking to use by compiling a cookbook. Recipes have been collected from friends and strangers. Cookbooks will soon be on sale and all proceeds will go to the Loeyes-Dietz Syndrome Foundation.

For more information, log on to Quentin’s Web site at www.quentinsquest.org.



Quentin’s check presentation to the LDSF

newlook

As you may have noticed, we’ve gotten a new look since our first issue. Our new logo is slowly making its way onto the scene along with new colors. New materials and an updated Web site will be along soon.

Be sure to check out www.loeyesdietz.org this fall/winter for the latest changes.

What do you share with your friends about LDS?

"It was in the past school year when I finally told one of my classes in speech form of LDS. I didn't really get to say much 'cause of the time limit and I guess I could of done it better, but my best friend and another good friend ended up asking me about it more afterwards. I never really wanted to tell my classmates about my syndrome or health problems cause I thought they would think I was some sort of contagious person.

During my speech it was actually rewarding, because when I looked at everyones faces I could tell that they were actually listening and not judging. Plus it was good to spread some awareness to other students.

I plan on doing more essays on LDS when I have the opportunity. My friends and other classmates are really nice though and they understand my limits and why I can't do certain things in gym and sports." - **Melissa**, 14 years-old

FUNdraising

FUNdraising can be FUN and easy for everyone involved! Often times, the hardest part is knowing where to begin. People generally want to make positive changes, but may find themselves asking how? The LDSF is putting together a FUNdraising Packet, and we would love to have your help and input in generating ideas to include in the packet.

Some of you have indicated that you would like to help with fundraising - here is your chance. Maybe you have experience with something that really worked, so just putting it down on paper will be simple! The idea of the FUNdraising Packet is to make fundraising easy and fun for all!

This packet will help identify successful fundraising ideas that will increase support for the LDSF, and build awareness.

Kids sometimes come up with ideas quicker and with more enthusiasm than adults. These lessons help children learn about community involvement and philanthropy, leadership skills, self-esteem, and how to work together toward a common goal! Two things that my children are involved with at their school is a penny war, with the highest giving class putting a pie in their teacher's face. Another is "hat day" where students pay to wear their favorite hat. What a fun way to get kids involved.

If you would like to help brainstorm ideas to be included in this packet, please email Beth at Beth.Utz@loeysdietz.org. I can't wait to hear all of your fantastic ideas.

baltimorecook-in

Thanks to all of our LDS families who joined us in the rain for a fun get-together this August in Baltimore! It was great to get to know everyone better, enjoying some great company and good food.



left: Alex, Gabi and Kelsey
bottom left: Carrie & Haley
bottom right: Enjoying photo ops



asktheexpert: Is there an association with LDS and cancer?

a: Mutations in the TGFBR genes have been associated with some forms of cancer. In LDS, only one copy of one of these genes is defective. In cancer cells, both copies of one of these genes has been damaged.

Importantly, it is not thought that inactivating the TGFbeta receptor is sufficient to cause cancer. Rather, this event can promote cancer progression if other cancer-related genes have also been altered.

While we have seen a few patients with LDS and a cancer history, this seems to be rare. It is unclear whether this is any different from what you would see just by chance. This issue requires further study.

- **Hal Dietz, MD.**

Questions for an LDS expert can be emailed to newsletter@loeysdietz.org.

contributions

The Loey-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.

We give a special thank you to those who gave in memory of Robert Hogan and in honor of Arthur and Dorothea Manning's 50th Wedding Anniversary.

All those who gave generously will be acknowledged at the end of the year.

holidaygiving

The Forever Gift

Thank you for considering a gift to the Loeyes-Dietz Syndrome Foundation. Please complete the donation form found at www.loeydietz.org, and return it, along with your gift to 4153 N US Highway 23, Fostoria, Ohio 44830. You may also make your gift online by clicking on the JustGive link, or by going to www.justgive.org and typing in Loeyes-Dietz Syndrome Foundation as your charity!

getinvolved

This fall/winter season, we are asking our LDS families to help the LDSF bring awareness to Loeyes-Dietz syndrome and help fundraise as we move forward to provide education, support and awareness about LDS. We have pins and notecards available for local resale with portions of proceeds going to the LDSF! Pins are made by artist Terri Kirchner and sell for \$15 with \$6.50 of each sale being tax-deductible and funding going to the LDSF. Notecards are also available for holiday cards/thank you cards with proceeds going to the LDSF! LDSF bracelets are also available.

Please consider buying greeting cards to use as personal cards or spreading the word to friends and families! Also, consider hosting a table at a local community event and selling LDS items. The Foundation is able to help provide you with LDS information for table tops! If you are interested, please contact Beth Utz at beth.utz@loeydietz.org and we will help get you the pins and or notecards! Look for more information soon on the LDSF Web site, www.loeydietz.org.

The LDSF is a registered non-profit corporation under the State of Ohio and is exempt from the United States Federal Income Taxes under section 501(c)(3) of the Internal Revenue System (EIN#65-1306978). All gifts are tax-deductible. We thank you in advance for your kind assistance and support.



raisingawareness

Thanks to Kelly for volunteering to tell her story and answer questions during the "clinical correlations" lecture at Johns Hopkins Hospital. First year medical students received a lecture from Dr. Dietz and asked questions of Kelly (LDS) and Carl (Marfan syndrome). It was a great opportunity to educate the students. We encourage other families to volunteer with their geneticist or cardiologist to help with medical education. It's a great and fulfilling way to bring awareness to LDS!

right: Kelly, Dr. Dietz and Carl



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