

# matters of heart

issue | volume 2 | spring 2009

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



LDS families enjoying a night out in Boston.

## founders notes

Thank you to everyone who has given generously of time, finances, support and advice to help support the

Loeys-Dietz Syndrome Foundation. We appreciate all that you have done and continue to do to help us achieve the LDSF mission. It's been incredibly rewarding (and fun!) getting to know many of you through different venues. For those who have indicated on your membership form an interest in volunteering, don't be surprised to hear from a committee leader. If you have talents or gifts that you'd like to put to good use, please don't hesitate to let me know!

In the meantime, we would love to get feedback, ideas, questions for experts, etc, to help fill our newsletter as we move forward ([info@loeysdietz.org](mailto:info@loeysdietz.org).) We do take your suggestions very seriously. For instance, on the forum, there was recent discussion about putting together an information packet on questions to consider when making a decision about cardiac surgery. What a great idea! Another helpful idea on our list of resources to develop for families includes a tracking form to follow the dimensions of aneurysms of the aortic

root and vasculature. Also, if there are events, activities or announcements that you would like to make to your LDSF family, please let us know. Lastly, if you have writing talents, we could definitely use your help!

Thanks to your continued support, we are able to further our research mission into LDS issues. We are now financially supporting the ongoing research of Dr. Anthony Guerrero, regarding the gastrointestinal issues of LDS. We are very proud and happy to engage physicians and researchers making a difference in our families' lives. Additional projects will be announced through the Foundation website and newsletter in the upcoming months. Because of our commitments to research and our goals to widen the resources and opportunities that we can provide to LDS families, we are likely going to introduce a membership fee in the upcoming year. This will allow us to cover administrative costs and help us keep up-to-date with mailing addresses. As you may have read in our annual report, we have come a long way and are excited about the path in front of us! So, keep checking back and supporting the LDSF.

Enjoy your spring!

Sincerely,

*Beth Utz and Gretchen Oswald*

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## foundation mission:

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

## raise funds

LDSF heart note cards and pins by artist Terri Kirchner continue to be available and make great gifts for Mother's day, Teacher gifts or any occasion! Cards are \$10 for a packet of 10 and pins are \$15, plus shipping and handling. Please email [Gretchen.Oswald@loeysdietz.org](mailto:Gretchen.Oswald@loeysdietz.org) for more information.



LDSF pins  
by Terri Kirchner

## 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.

**Loeys·Dietz**  
Syndrome Foundation

“It’s been a hard journey but everything is different now that we found the answer and aren’t alone.”

- Sheila, LDSF Board Member



## heartrenderings

*Raquel Gomes is a Board Member of the LDSF and mother to 6 year old Gabi, who has LDS. The following is part of Raquel’s reflection of her family’s journey with LDS.*

Second only to motherhood, LDS has been a mixed source of distress and sheer celebration of life. My six year-old daughter Gabi was diagnosed with LDS in early 2005. At three, Gabi had already had cranial reconstruction for craniosynostosis and eye muscle surgery to correct for “lazy eye;” severe food allergies and constant abdominal cramps and bleeding; and about a year’s delay in her gross motor

skills. She was also on Propranolol for her dilated aortic root, for which we knew she’d eventually need surgery, most likely in adolescence. Getting the diagnosis of LDS – her first diagnosis for which there was an actual genetic marker – in itself wasn’t that life-changing. At first.

Within a year and half of her diagnosis, we learned that LDS meant much earlier intervention to correct the dilation. Waiting for the 5.0 cm threshold was no longer an option. Gabi had her aortic root repair in late 2006 and, almost inexplicably, became another child. Within months of surgery, her food allergies subsided, her nights became sounder, and her energy and stamina were like never before. Instead of retreating to her favorite corner of the sofa as was the norm, here she was dancing through the house with her sparkly wand in hand. The right diagnosis became life-changing.

At this point I’ve been through the many phases of any parent whose child has a medical condition: despair over the unexpected, mourning over the loss of “perfection,” to gradual acceptance, and finally, the bliss of enjoying the mundane in life despite it all. No doubt it’s taken its toll, in too many sleepless nights, grey hairs, and marital stress. But it’s also been a matchless reminder of what really matters in life.

Yes, I still worry about aggressive aneurysms, arterial tortuosity, possible dissections, and recurring headaches (not just about Tylenol for a child with hydrocephalus). I also worry about her self-esteem and her view on life, as she becomes increasingly self-aware in a world of gawking stares and painful preconceptions.

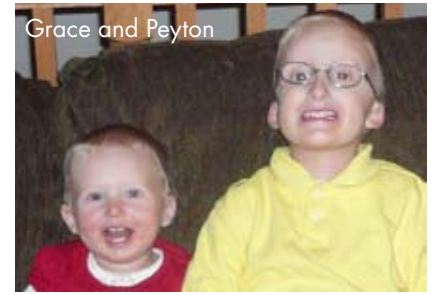
For the most part though, I’m now living in a state of grace. Gabi’s health is stable, she’s thriving, happily enjoying life in more ways similar to other kids. There are even those days I forget we live with LDS. And life is bliss.

At the same time, given that LDS is part of our lives, my state of grace also extends to the privilege of counting on Dr. Dietz, Dr. Loeys, Dr. Cameron, Dr. Guerrero, Dr. Slack, and the many other experts dedicating their minds, and hands, and hearts to understanding more about LDS. If Gabi and others with LDS are able to lead more full lives, it’s in no small part because of them.

Likewise, the blessing of being a part of the LDSF community. There’s the comfort of sharing a common language. There’s the amazement in witnessing others’ commitment to the Foundation, their many journeys, and most welcome sense of humor. And there’s the hope that together we provide helpful resources for newly diagnosed patients and their families, raise awareness about LDS, and support research efforts that will benefit our growing community.



Gabi and Raquel



*Hi, my name is Peyton and I am a 3 year old boy. I live in Hobart, Indiana, with my parents and my baby sister, Grace. I love to play Super heroes, pirates, police officer and bad guys, and video games. I love to play and watch sports too; even though I can’t play most of them. Every day is different for me. I see a lot of different doctors and I take medicine every day for my heart. I have Loeys-Dietz Syndrome (LDS). I am not the only one with this disease; my dad and one year old sister have LDS too. This is why we need your help. We are bowling for a cure. Find out more in the Upcoming Events section.*

## inlovingmemory:

### Erin Squires Ramsey

November 22, 1992- December 25, 2008  
Puyallup, Washington

Erin was a sophomore honors student at Emerald Ridge High School. She enjoyed spending time with her friends, reading, fencing when her health allowed it, and verbally jousting with friends and family. Erin had a light, strength and sense of self beyond her years.

*Our fondest thoughts and well wishes go out to the Ramsey’s family and friends.*



## contributions

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.

# connective tissue disorder awareness group

We congratulate and support LDSF member Rachel Martin for her work in bringing attention to connective tissue disorders in Ireland, and hopefully beyond. We asked Rachel to introduce herself, co-leaders and group to the LDSF community.

The Connective Tissue Disorder Awareness Group has been set up by Rachel Martin, Lisa Fagan and Wendy Rossiter; all from Dublin, Ireland.

We originally called our group the Connective Tissue Disorder Action group but feel that "Awareness" is possibly more appropriate for what we hope to achieve. We are all passionate about achieving greater public awareness about connective tissue disorders. **So, who are we?**

**Rachel Martin:** Rachel was born in 1980 and was a noticeably floppy baby. In infancy, she was noted to have a cleft palate and "split" uvula. At 12 months of age, Rachel had a diaphragmatic hernia and hiatus hernia repaired. At 18 months, she underwent cleft palate repair. Numerous cosmetic surgeries were performed during childhood on her feet and legs as well as pectus carinatum repair. Scoliosis appeared during the 1990's. Up to this point Rachel was told she had congenital myopathy. Her pediatrician had mentioned Marfan syndrome during earlier hospitalizations, but there were no heart problems at that stage, so it was ruled out. In 1999, she was clinically diagnosed with Marfan syndrome after aortic enlargement was detected. In July, 2008 at the National Marfan Foundation annual conference in Boston, Massachusetts, Rachel was officially diagnosed with Loeys-Dietz syndrome, which was later confirmed by genetic testing. She was advised by geneticists Dr. Milewicz and Dr. Dietz to have heart surgery as a matter of urgency as her aortic root was at 4.1 cm. She returned to Ireland, and after lengthy discussions with cardiothoracic surgeon, Professor J. Mark Redmond, had valve-sparing aortic root replacement surgery on September 9, 2008.

**Lisa Fagan:** Lisa was born in 1980. Throughout childhood she had many visits to local children's hospital for detached retinas and had several operations to repair. Lisa is tall and slim with long digits and scoliosis. Marfan syndrome was never officially diagnosed. More recently, Lisa also had numerous medical complaints which she continuously raised with her doctor, but felt that these concerns were never taken terribly seriously. After some personal research, Lisa questioned the diagnosis of Marfan syndrome and requested a referral to a cardiologist. She saw cardiologist, Dr Kevin Walsh in July, 2008 and was clinically diagnosed with Marfan syndrome. Thankfully, to date, Lisa's aorta has not required surgery.

**Wendy Rossiter:** Wendy is the mother of Martin, who sadly passed away in July 2008. Martin was born in 1979. In 2004 he was diagnosed with Marfan syndrome, despite having many symptoms noted earlier in life including tall stature, long limbs, and scoliosis. He was a successful

basketball player for Ireland. Unfortunately, at diagnosis he had to give up his basketball career. At 23, he suffered an aortic dissection, was monitored in the ICU for a number of days and then discharged on medication. No surgery was performed or planned. He then moved to the UK where he underwent surgery to repair the dissection and an aneurysm in his abdomen. Two years later Martin was told that his entire aorta would need to be replaced. The doctors in Ireland were unable to perform the operation as it had never been carried out before and carried a high risk of death or paraplegia. Martin made the decision to travel to the UK to have the surgery. The surgery took place on July 10, 2008 (the same date Rachel was diagnosed with LDS) and although the surgery itself was successful, complications following the surgery led to Martin sadly passing away. He has three children, two of whom have Marfan syndrome and are being closely monitored.

As we have all had a difficult time with getting correct diagnoses of the genetic conditions affecting ourselves or our family members, we feel that it is time that the public were made more aware of the seriousness of misdiagnoses of connective tissue disorders. Aches and pains aren't always just in a person's head; often they are rarely further investigated. Unfortunately, in many instances, it relies on a tragedy or emergency for the correct diagnoses to be given.

Our group's goal is to raise awareness with the medical profession, the public, and sports teachers in schools (a majority of European schools have sports as a compulsory subject.) We believe that the public needs to be more aware of their bodies and push towards medical investigations and diagnoses of connective tissue disorders.

We will be approaching various media outlets to have stories published and bring awareness to our group of disorders. We all feel very passionate about making information about connective tissue disorders publicly known.

Of course we will also be offering support in any way we can to those who have questions or are diagnosed. We will be working alongside different Marfan syndrome support groups, the Loeys-Dietz Syndrome Foundation and the various other support groups for other syndromes. It is our hope that any funds that we have donated and/or raised will be divided between each of the groups to be used towards research into the various conditions.

We invite all on Facebook to join our group – Connective Tissue Disorder (CTD) Action Group. We also have a temporary e-mail address [ctdactiongroup@gmail.com](mailto:ctdactiongroup@gmail.com). A website will be developed hopefully in the very near future.

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## asktheexpert: My test results say that this exact gene mutation has not been described before, is this common? Does it still mean I have LDS?

**a:** If the lab reports that a gene mutation has not been described prior, one needs to consider two possible alternatives. First it could be that this "mutation" is a so-called rare normal variant or polymorphism with low significance or secondly, it is a real bonafide mutation with high clinical significance.

In order to distinguish these two situations several things can be done. If the patient is the only one in the family to have clinical features of LDS, testing of parents can be performed to prove that the "genetic alteration" is not present in either of them and as such is a "de novo" or new mutation. This is a strong argument for causality. If the patient is part of

a family with several affected individuals, it is very informative to check for the presence of the genetic alteration in several individuals.

If all affected individuals have the mutation and none of the unaffected carry the change, this is again a strong argument for the causality. Finally, depending of the location, type and nature of the specific gene mutation, one can feel more or less confident about causality. The latter can be discussed with a geneticist familiar with the gene and the disease.

**- Bart Loeys, MD.**

Questions for an LDS expert can be emailed to [newsletter@loeysdietz.org](mailto:newsletter@loeysdietz.org).

# upcomingevents

## Pins For Peyton

Saturday, April 18th  
Hobart, Indiana

Join the Griffith family and have bowling fun while knocking down some "Pins for Peyton!" (And for those who don't bowl, there will be Texas Hold 'Em fun in the lounge!) Following is an excerpt from Peyton. For his full letter or more information please check out [www.pinsforpeyton.com](http://www.pinsforpeyton.com) or the "Pins for Peyton" page on Facebook!"

## Crawfish Boil

Saturday, April 25th  
Fort Worth, Texas

The Bilbow family is preparing for the 2nd Annual Crawfish Boil in Texas. If you are in the area and looking for a great afternoon of music, beer and plenty of crawfish, or would like to help out, please contact [kendallandjohn@sbcglobal.net](mailto:kendallandjohn@sbcglobal.net)

## Quentin's Quest, Part II

Quentin Waid's cookbook to benefit the LDSF is coming along smoothly, with about 300 recipes collected from LDS friends and families around the world. More details to come this spring.

## Ohio Golf Outing

Saturday, September 19

Fallen Timbers Fairways, near Toledo Ohio  
The Utz family is working with local businesses and families for a golf outing this Fall in honor of their son, Alex and the LDSF. Contact Beth ([Beth.Utz@loeysdietz.org](mailto:Beth.Utz@loeysdietz.org)) for more details or to help out.

**Loeys-Dietz**   
Syndrome Foundation

4153 North US Highway 23 Fostoria, Ohio 44830  
[www.loeysdietz.org](http://www.loeysdietz.org)

# springFUNdraising

## April showers bring...May Bargains! May Madness Garage Sales to Benefit the LDSF

Help raise awareness for Loeys-Dietz Syndrome one garage sale at a time  
How Do I Help? Is it really that easy?

- Yes! Pick a day(s) in May to have your sale!
- Ask neighbors, family members, coworkers to donate items (and volunteer to bring whatever doesn't sell to a donation site!)
- Contact the LDSF for educational material to set up at your sale to get the word out about LDS.
- Donate proceeds to the Loeys-Dietz Syndrome Foundation, 4153 North US Highway 23, Fostoria, Ohio 44830

Any questions may be directed to [Beth.Utz@loeysdietz.org](mailto:Beth.Utz@loeysdietz.org) or [Barbara.Hawthorne@loeysdietz.org](mailto:Barbara.Hawthorne@loeysdietz.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.*

## globalfriends

We welcome all of our families from around the world, including Tristan from Germany! Other countries represented on the LDSF Forum include Australia, Belgium, Canada, England, Ireland, Japan, Netherlands, New Zealand, Scotland and Singapore.



above: Tristan

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