

matters of heart

issue2 volume2 summer2009

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



Beth Utz (left) and Gretchen Oswald (right) show off their pirating skills in Baltimore's Inner Harbor

founders notes

On June 26th-27th, some members of our LDS family truly experienced the power of community in Baltimore. In

conjunction with clinic appointments, more than 60 family members came together for an informal discussion with LDS specialists at Johns Hopkins, followed by an Orioles Game with fireworks and a Pirate Ship Ride!

Thank you to Carolyn Dinsmore and Laura Hutchins who helped out watching kids and for all the families who

came together. A special thank you to Chip Weinman and Family (Skybox O'ssociates), Gabi Tapkas, Coordinator of Corporate Sponsorship & Suite Services of the Baltimore Orioles and ARAMARK for their generous donations of suites, tickets, snacks and O's tee shirts! It was great to see the LDSF recognized on the scoreboard!

As the LDSF continues to grow, we are striving to create larger events for families to come together, get new research and treatment information, and share their stories. (Oh, yes, and have a lot of fun!) Stay tuned for more information on how you can get involved.

Sincerely,

Beth Utz and Gretchen Oswald

fun in baltimore

"I feel that Mercedes has come away with friends with which she has a very deep connection. She is nine yrs old, and really needed the contact with other LDS children so as not to feel "different" from every other child. We live in a very small town and she knows no other children with health issues... not even asthma. She has been jealous of her friends "because all they have to do to get ready is brush their teeth!" She now counts amongst her friends, kids that have a similar life to hers. She is no longer alone in her differences." - Eden, mother of LDS child



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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

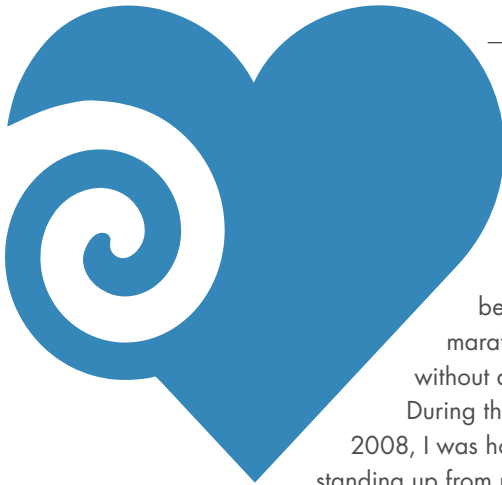
matters of heart mission:

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

"I liked meeting kids that were different, like me surgeries...crooked fingers...braces they understand what happens to me and how horrible surgeries are.."

- Mercedes, age 9, LDSF Member on meeting other LDSF families in Baltimore



heartrenderings

Federico Brugnoli of Italy was diagnosed with Loey's-Dietz syndrome in 2008. He shares his amazing story of perseverance and optimism. His story also highlights the importance of early diagnosis and treatment.

My health has always been perfect. I ran marathons and played rugby without any problems. During the night of April 5th, 2008, I was home alone and after standing up from my chair I felt a chest pain; everything went gray. After fainting, I awoke to a stabbing chest pain and was unable to move my right arm and leg. I had never fainted before, so I thought everything would be ok. I still don't know where that strength came from, but I stood up, drank an energy drink and called my parents. The situation didn't seem to get better, so I called for an ambulance. My sight started to get worse and I was scared. I would never have called "911," preferring to "wait and see," but that pain wasn't normal.

I was able to knock on my neighbors door and they helped talk me through my fears and anxiety. The ambulance arrived and shortly after getting to the hospital I was able to move my right side again. After a few exams, the doctors said that they would continue investigations in the morning since my situation didn't seem threatening. They told me to sleep and for my parents to go home.

My safety, my angel, the man I will thank for the rest of my life, was an Anesthetist who asked himself whether there were any problems with my heart/aorta. He woke the Cardiologist and imaging revealed an aortic dissection in my ascending aorta. My parents had returned home and were not able to make it back to the hospital before I went in for surgery.

After 3 days in the hospital I could walk a little bit and I started to feel better and better. Soon, I was able to be on my own again and to follow the lectures at my university.

My first CT scans post surgery were scheduled for July. My doctors discovered that my abdominal aorta was dissected, and it was about 6.5 centimeters wide. They told me they would have to perform another surgery, but not for another 10 years. The next CT scan was scheduled for January 2009. I felt quite ok, but my mother didn't...

The point is that nobody ever dared to ask themselves "why did all this happen to a 20 year old boy?"

My mother saved my life. She found a research center for genetic cardiovascular diseases in Pavia, Italy. Professor Arbustini, head of the research center, diagnosed me with LDS and after further imaging she discovered my abdominal aorta was much worse than the former

doctors thought. A surgery had to be performed as soon as possible. She found Professor Odero, who had enough guts to perform it. My future was to be decided on October 15, 2008.

I was quite worried, but also sure that my case was followed by some of the best professors in Europe. After taking my last exams in September, I was ready. No fear at all.

What my parents told me about that day is terrible. The surgeons found the situation much worse than expected. My aorta was around 9 centimeters wide and very thin; they told me they could see through it. Surgery took 16 hours with many complications. But I survived (and so did my parents!)

The following two weeks in the NICU were rough. I wasn't able to move my legs and I ended up having a tracheotomy. I couldn't talk at all. It was such a pleasure to see my family again, even without being able to talk. It was a long recovery. I lost 45 pounds and had problems with vocal cord paralyzation. This is still really difficult because I had loved singing with my band.

Further imaging revealed another aneurysm which required multiple surgeries, which went well. My eating problems have resolved and I've had more energy.

During all these months, my mother left her job and rented a flat next to the hospital. My father of course had to work, but he was visiting us every weekend. They were so strong!

In March, 2009 I was moved to a clinic close to my city so my family moved back home. I regained my weight, and I became quite independent in my wheelchair. On June 5th, I finally came back home, after 8 crazy months! Nobody knows for sure if I will ever be able to walk again, but I am so happy to be alive, and I will always keep on fighting and hoping. I am with you!



Federico Brugnoli

contributions

The Loey's-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.



Pins For Peyton

Hobart, Indiana

Congratulations to Peyton and the Griffith family for their "Pins for Peyton" bowling event on April 18th! With bowling and raffles, **over \$15,000 was raised!**



Garage Sale

Fostoria, Ohio

Thank you to the Buckeye Clover Bunch 4-H group in Ohio for organizing a garage sale for their community service project, with all proceeds to benefit the LDSF. The group **raised over \$600** selling items, face painting, lemonade stand and cookies, all while spreading awareness for Loey's-Dietz Syndrome!



Bike Run and Spaghetti Dinner

Plum, Pennsylvania

Jacob Knighton is a thirteen year old boy diagnosed with Loey's-Dietz syndrome, after experiencing an aortic dissection on New Years Eve in 2005. Children's Hospital in Pittsburgh performed emergency surgery replacing the aortic valve and root. In April 2009, Jacob experienced another dissection of his aortic arch and required a second emergency surgery. Although it has been difficult for Jacob to not play sports, he remains upbeat and active, with great support from his community.

On July 16, members of the community came together for Bike Run and Spaghetti Dinner. More than 100 bikes took part! Michelle Knighton, Jacob's mother reports, "It was awesome! It was a huge success and we had a great turn out! People enjoyed the dinner and were very generous in the raffle and auction. We put flyers out with information on Loey's Dietz along with the web site. I am hoping that we brought some awareness to our community."



Crawfish Boil

Saturday, April 25th
Fort Worth, Texas

Thank you to the Bilbow Family in Fort Worth, TX for throwing the Second Annual LDSF "Crawfest" on Saturday, April 25.

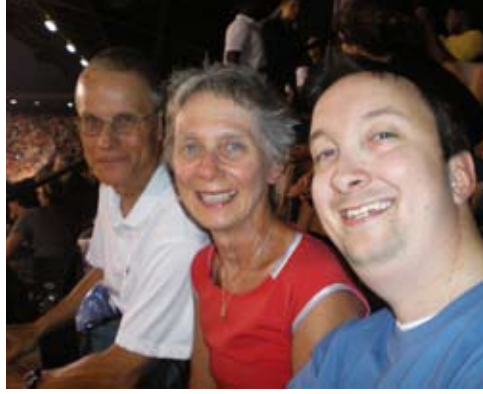
As you consider your own fundraising, consider the words of Kendal Bilbow, mother to 6 year old Jylian.

"As the mother of a very special child who has LDS, I'm often lost for words as I try to explain the emotional as well as physical impact that Loey's Dietz Syndrome has had on our lives. Living with LDS can be a scary, sometimes isolating experience, but we love and cherish every day as we diligently pursue new treatments and support for our precious Jylian (Jelly Bean)! Please commit to being a part of fundraising for the LDSF. Your life will be impacted as you play a vital role in impacting our LDS community!"



more fun in baltimore

"It was a wonderful chance to spend time with other families that share in the same LDS 'Foot Print.' I am very excited to attend future gatherings." - Stacey, mother of LDS child



raisefunds

LDSF heart note cards and pins by artist Terri Kerchner 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 for more information.

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-deductable. We thank you in advance for your kind assistance and support.



LDSF pins
by Terri Kirchner

Loeys·Dietz 
Syndrome Foundation

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