# *Channels*

News, Events & Features from the Lymphangiomatosis & Gorham's Disease Alliance

"Reaching for a Cure"
Winter 2009


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## PRESIDENT'S COLUMN

Welcome!

Indeed, a warm welcome to the first issue of Channels, the newsletter of the Lymphangiomatosis & Gorham's Disease Alliance. The purpose of Channels is to provide periodic reporting to our community of patients and their families, and to all those who are interested in the activity of the LGD Alliance. Our mission is to improve the care of patients with lymphangiomatosis and Gorham's disease by being a force for research that will find effective treatments and a cure for these diseases. We have so many people to thank who have helped us form the LGD Alliance. But first, please take the time to read through this issue's feature stories and news updates.

The LGD Alliance was founded in July 2007. We had investigated and studied many of the other orphan-disease foundation organizational models. We are so grateful to all for their insight - especially those founders who met with us personally. We recruited our first Board of Directors, our Medical Advisory Council, and held our first Board Meeting in August. Soon after, our website was designed, implemented, and became accessible via the Internet. In the fall of 2007, we established the first-ever directory of Doctors Around the World (DAW's) - physicians & surgeons in every area of medicine who have seen and treated or consulted in the case of a patient with either Gorham's or lymphangiomatosis. We commend all the doctors who have consented to be listed in the DAW's directory. By year's end, we had met with the staff of the Vascular Anomalies Center at Children's Hospital Boston, and established a formal referral procedure for case review, evaluation, and appointment requests. Children's Hospital Boston became our first designated multidisciplinary VAC. In December, we elected three new Directors, increasing Board membership to seven.

In 2008, our first full year, we focused on establishing a worldwide patient network of Gorham's and lymphangiomatosis. In support of this goal, we upgraded our website to add a Patient Forum and established a Patient Directory - as an important first step towards developing a Patient Registry. We have recently begun discussions with a University Medical Center to define our requirements for a Registry. To extend tax advantage consideration to donors to the Alliance, we applied for and received, in the first quarter, our certification from the IRS designating the foundation as a 501(c)(3) public charity. In connection with our goals to develop a research plan and to spread awareness effectively among the public and medical/scientific communities, we established membership in the National

Organization of Rare Disorders (NORD), the Genetic Alliance, and the Rare Bone Disease Patient Network (RBDPN), which is an entity of rare bone disorders created under the auspices of the United States Bone & Joint Decade (USB&JD).

Further, in June, we received an invitation to, and attended, the 17th International Workshop on Vascular Anomalies at Harvard Medical School, and the International Society for the Study of Vascular Anomalies (ISSVA) 2008 conference. Also in June, we attended, by invitation only, the Bone Summit in Washington D.C., which was convened by the National Coalition for Osteoporosis and Related Bone Diseases to develop and make recommendations for a national bone-health action plan. Participating were more than 150 individuals representing an array of stakeholders concerned with bone health. Lead attendees included the U.S. Surgeon General and Congressman Patrick Kennedy of R.I. In October, along with our fellow members of the RBDPN, we participated in the historic 1st Advances in Rare Bone Diseases Scientific Conference at the NIH in Bethesda, MD. Along with our RBDPN colleagues we participated in the American Society for Bone & Mineral Research (ASBMR) Conference in Montreal. These external activities have helped greatly in spreading awareness as well as in connecting with many more medical specialists, investigators, and clinicians and researchers who have some knowledge and/or experience with lymphangiomatosis & Gorham's disease - or are willing to help in our efforts in some meaningful capacity.

These are our first steps on the journey to find the markers that help find the cure. We don't yet know how distant the day is when we can begin to see the long dreamed-for treatments. We have much to do - but we have begun. We shall strive to find the science tracks which accelerate the searches and find the causes. We will focus on raising the funds needed to attract investigators who will be charged with conducting this research.

In 2009, we will be aiming to achieve the commencement of the first-ever scientific research programs that will lead to the development and trials of effective treatments. We need your help on the journey. We hope you will come along. We express our very sincere appreciation to all those who have helped in the starting phase; please see the special list in this newsletter.

Jack Kelly

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## FEATURE STORY

Jana Sheets - Founder

As the Founder of the LGD Alliance, Jana Sheets is a pioneer in the struggling effort to finding a cure for two orphan diseases - lymphangiomatosis & Gorham's disease. Jana has lived with lymphangiomatosis for nearly 30 years and aside from the love and support provided by her family and friends, and several very special doctors, her primary source of strength lies within the fact that she is her own expert.

After graduating from high school, and with the growth of information on the Internet, Jana started using her mom's computer to conduct an unwavering search for information on her

condition and to try to find someone else in the world with her disease. Throughout her whole life, she'd always asked her doctors if they had other patients like her or if they'd ever even seen another person with lymphangiomatosis. The answer was always something like "you are a needle in a haystack", or "I could live to be 1,000 and would never see another patient like you". It took years of searching and searching before she stumbled upon a personal website created by the Petersen family from Denmark. The site was dedicated to their son, Marcus, and his experience with Gorham's disease and lymphangiomatosis. It had a message board and it was there that Jana finally connected. It was there that she also realized the need for something bigger. This small worldwide orphan-disease community urgently needed help and she intended to do something about it.

Her plans were disrupted when her illness took a serious turn and she was told she would need a bi-lateral lung transplant to save her life. She and her husband, Eric, decided to immediately move to Durham, NC so that she could have her life-saving surgery at Duke Medical Center. In May 2004 she received her new lungs and with that made medical history as the first patient with lymphangiomatosis to successfully undergo a bi-lateral lung transplant. As a result, her medical team co-authored a case report that was published in the American Journal of Transplantation. This paper would allow for the knowledge gained from her surgery to be shared with the medical community and give the possibility of transplant to other patients with end-stage lung disease resulting from lymphangiomatosis. Because of Jana's extensive knowledge about her condition, her team of doctors invited her to be a co-author of the report.

After enduring a long and rough recovery, Jana felt more than ever that she had a purpose, a calling she could not deny. She was an adult who'd survived a rare pediatric illness - a feat that all of her doctors had told her parents was unlikely. She'd now gone on to survive again - against all odds. She had a wealth of information and experience which she wanted to share with other patients and caregivers. She wanted to share the journey she had traveled and felt that her story would provide encouragement and hope.

In July 2007, the LGD Alliance was established by Jana, her father Jack Kelly and a few families and patients with the 2 conditions. The website, www.lgdalliance.org, went live in January 2008. It became a beacon for patients and caregivers and from the moment it was accessible over the worldwide web, the LGD Alliance began fielding inquiries from people all over the globe. As the Director of Patient Support, Jana devotes her time to answering every new inquiry, responding to all requests, helping people find doctors, connecting them with other families, and offering them hope. It is her belief that being an informed patient and caregiver is the key to survival. She has learned over the years the sad fact that she's more knowledgeable about her condition than most medical professionals. It is because of this that she advises everyone to learn as much as they can about the diseases. The patients who turn to her seeking help and support continuously inspire Jana. In return, she is their source of confidence and inspiration.

Jana hopes that in the coming year, the LGD Alliance will have raised the funds to allow them to put their research agenda into motion. In addition she also continues on her quest to convince scientific investigators, medical researchers, and institutions of the need to understand and study lymphangiomatosis & Gorham's disease. This is the mission of the LGD Alliance and thanks to Jana Sheets, improved treatments and a cure are getting a little closer to reach.

# FUNDRAISING UPDATES

## Funding Research

The primary work of the Lymphangiomatosis & Gorham's Disease Alliance is to support our patient community and to organize an effective, accelerated, science-based research program. Foundation resources to support these objectives come from an all-volunteer cadre of people - some of whom work full-time, many who devote their precious off-hours to help out. Presently, there are no paid employees or staff, nor has compensation of any kind been paid to officers or Board members.

The Officers and Board of the LGD Alliance are currently developing our initial Research Plan. In this connection, our Research Committee has undertaken the responsibility of forming our Scientific Advisory Board. Our 2009 goal is to fund the first-ever scientific investigations to define the natural history of lymphangiomatosis and Gorham, and after a proposal-selection process, to award the first projects to search for the causes. These steps take us onward in the journey to testing developing therapies, organizing patient trials and hopefully delivering effective treatments. To accomplish this we need to raise funds. Our revenue development plan is being formulated to support these research projects. Our research vision is aggressive; our fundraising target is sizable. Details about the projects being contemplated and the funds needed to activate them will be forthcoming soon on our website and in the forthcoming Channels.

The state of research looking into the causes of lymphangiomatosis or Gorham's disease is indeterminate, imprecise. A study on the natural history of either Gorham or lymphangiomatosis is nowhere to be found. Throughout the medical literature there is controversy even as to what name to call these two very serious orphan diseases. As to treatments, there are no standards. As was recently expressed at the 1st Advances in Rare Bone Diseases Research - "In Gorham's we are at the beginning - building a knowledge base about the causes".

We know very little about the causes - but we are going to change that - if you come along on the journey. Our patients need your help - and the need is now. Please let us know if you can help - even with the smallest amount. Every single dollar will advance the journey to that long dreamed-for cure. Lymphangiomatosis and Gorham's disease have taken so many lives of adults, children and young adults. Several patients who succumbed in 2007-08 had expressed their last wish by having donated their bone tissue and serum for research. One young person, age 11, said - I give my body in the hope that no child ever has to go through what I have gone through.

Please give to support the research.

## Research News

LGD Alliance Participates in First Scientific Conference
Advances in Rare Bone Disease
October 22-24, 2008
National Institutes of Health - Bethesda, MD


In early 2008, the Lymphangiomatosis & Gorham's Disease Alliance (LGD Alliance) joined the Rare Bone Disease Patient Network (RBDPN) - an entity created under the auspices of the United States Bone & Joint Decade (USB&JD) to expand research on rare bone diseases, foster information exchange and idea sharing, increase rare bone disease awareness, and advocate for government policy and funding in support of rare bone, and rare bone-related, diseases.

To step up the interest by the research community, a first-ever conference was planned in 2007 to gather scientists and medical professionals with an interest in rare bone disorders to present their work in support of rare bone disease research. A committee of medical, scientific and foundation executives planned and organized the 1st Advances in Rare Bone Diseases Conference which was recently held at the National Institutes of Health (NIH) in late October 2008. The meeting was a special milestone as the first-ever gathering together of scientists, clinical researchers, and patients involving several serious bone disorders.

The Network member organizations, in addition to the Lymphangiomatosis & Gorham's Disease Alliance include: - Fibrous Dysplasia, Fibrodysplasia Ossificans Progressiva, Melorheostosis, Osteogenesis Imperfecta, Osteopetrosis, Paget's Disease of Bone and Related Disorders, and XLH - Rickets. The international gathering provided an exchange and sharing of knowledge through plenary lectures in genetics, molecular therapy and drug development for orphan diseases. Young Investigators presented recent work in the specific rare disease areas. Important state-of-the-art presentations were given by a number of noted world specialists in bone biology, molecular therapy, nano technology, development of the skeleton, nervous system control of skeletal diseases, and on the base science of osteocytes.

The high point for us was the first-ever Gorham's disease & lymphangiomatosis presentation that we were invited to make to this deeply talented gathering of researchers. Our disease profile was made by John Reith, MD, Director, Bone & Soft Tissue Pathology, Associate Professor, Pathology, Immunology and Laboratory Medicine, University of Florida Medical School. Dr. Reith is a founding member of the LGD Alliance Medical Advisory Council. Dr. Reith's presentation was historic - the first made to such a special gathering of scientists and clinical researchers on behalf of the community of patients of Gorham's disease and lymphangiomatosis the world over!

During this 1st Advances in Rare Bone Diseases Conference, there was a special breakout - "fireside chat" - session for patients, advocates and foundation representatives to talk with a scientific/clinical research panel with knowledge or interest in lymphangiomatosis & Gorham's disease. The professional panel included Dr. Reith, Jay Janicki, MD, an orthopaedic surgeon from Chicago, and Dean Bjorn Olsen, MD, PhD, distinguished professor of Cell Biology at

Harvard Medical School. This was another historic moment for the LGD Alliance: the first time, ever, a group of patients/families, had an opportunity to sit down and meet with medical and scientific professionals to talk about the desperate need to engage appropriate resources to launch a formal research effort to define the natural history of our disease(s) and to assist in the development of an effective research plan.

In our journey to find better treatments which save and improve the lives of patients afflicted with lymphangiomatosis & Gorham's disease, we can mark the days of October 22-24, 2008 as a milestone - as a time when the journey not only advanced - but rose to a new level. The journey continues.

We would like to express our sincere appreciation to all of our co-members of the Rare Bone Disease Patient Network (RBN), the USB&JD, the members of the special planning committee, and funding supporters, including the National Institutes of Health (NIH), the American Society for Bone & Mineral Research (ASBMR), BioMarin Pharmaceutical, Inc, Enobia Pharma, Inc, Genzyme Corporation, The Hope & Grace Foundation, Kyowa Hakko Kirin Company, LTD, the Orthopedic Research Society (ORS), and the Weldon Golf Tournament, Many thanks to all those who attended, and our special gratitude for those who presented. We are grateful to Charlene Waldman, (Paget's Foundation), Toby King, (USB&JD), Emily Clarke, (USB&JD), Kathleen Harper, (Melorheostosis Foundation), Hillary Weldon, (IFOPA/USB&JD), and Michael Econs, MD & Craig Langman, MD - co chairs. And, surely to John Reith, MD who carried our banner! All gave so much of their time and skills towards making this meeting such an outstanding event.

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## EVENTS

Rare Disease Day - Feb. 28th

To celebrate the Second Annual Rare Disease Day, the LGD Alliance will join as a partner along with hundreds of other patient organizations, government agencies, medical societies and companies in focusing attention on rare diseases on that day. This day aims to raise awareness of rare diseases, the special challenges encountered by those affected, and the need for research to develop safe, effective treatments or cures. As a partner, the LGD Alliance will help promote Rare Disease Day and members are encouraged to publicize the day as much as possible, including writing a letter to the Governor to request the last day in February be designated Rare Disease Day in the state.

# BOOK OF HONOR

Thank you all so very much.  You are dear friends and we love you.

| *Contributor* | *Remembering* | *Honoring* |
|---|---|---|
| Marjorie A. Ravas | | Jana K. Sheets |
| Jana K. Sheets | | Patient Community |
| Wendy & Bennet Chaite | | |
| Kathleen Olson | | |
| Eric & Jana Sheets | | Jack Kelly's Birthday |
| Carol & Jack Kelly | | |
| Sandy & Scott Goldfarb | | |
| Allan Gammon | | |
| Jack Kelly | | |
| Suzanne E. Fitzsimmons | | Jana K. Sheets |
| Sandy & Scott Goldfarb | | |
| Sandy & Scott Goldfarb | | |
| Anonymous | | Jana K. Sheets |
| Gina Bosco | | Zachary Goldfarb |
| Renee Nickerson | Robin Lee Brown | |
| Annette & Robert Petersen | Marcus Petersen | |
| Gavin Sheets | | Jana K. Sheets |
| Donald Ray & Alice Simpson | Sonia Herbert-Brande | |
| Brittany Ferguson | Sonia Herbert-Brande | |
| Carolyn McCrudden | Sonia Herbert-Brande | |
| Diana & Harold Christian | Sonia Herbert-Brande | |
| Jack Kelly | Sonia Herbert-Brande | |
| Gina Bosco | | Zachary Goldfarb |

| | |
|---|---|
| Leigh F. O'Callaghan | Sonia Herbert-Brande |
| English Construction Company | Sonia Herbert-Brande |
| Nell & Perry Frye | Sonia Herbert-Brande |
| Pamela Anne Bell | Sonia Herbert-Brande |
| Mary Devon McWilliams | Sonia Herbert-Brande |
| Vern & Helen Roberts | |

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On behalf of the worldwide community of children, young adults, and adults with lymphangiomatosis or Gorham's disease, The Alliance wishes to convey our deepest and whole-hearted gratitude to all those listed below. You shall always be remembered for helping us take the first steps on our journey searching for better health.

## Founder

Jana Sheets

## Board of Directors

| | | | |
|---|---|---|---|
| Allan Gammon | Shannon Gareau | Sandra Goldfarb | |
| Sonia Herbert-Brande* | Jack Kelly | Kathleen Olson | Scot Wiesner |

*Sonia Herbert-Brande, a Founding Director, passed away December 28, 2008.

## Medical Advisory Council

| | | |
|---|---|---|
| Denise Adams, MD | Ernest (Chappie) Conrad, MD | Timothy A. Damron, MD |
| Jerry P. Eu, MD | Steven J. Fishman, MD | Teri J. Franks, MD |
| Paula E. North, MD, PhD | M. Anthony Pogrel, DDS, MD | John D. Reith, MD |
| Mark Scarborough, MD | Thomas C. Shives, MD | Michael P. Whyte, MD |
| Mark E. Wylam, MD | | |

## Family, Friends, Companies, Institutions & Partners

AccountAbility Specialists, Inc. - West Palm Beach, FL

Stuart Ames, Esq. - Miami, FL

Asociacion de Lucha Contra La Enfermedad de Gorham (A.L.E.G)

Rhonda Brown - Latrobe, PA

Linda Buettner - Greensboro, NC

Bluewave Graphics - Delray Beach, FL

Sue Byrnes - Founder, LAM Foundation

Wendy Chaite, Esq. - Founder, Lymphatic Research FoundationCommunity Foundation of Palm Beach & Martin Counties, FL

DAW's - The doctors who've consented to be in our referral directory

John Dixon - Durham, NH

Duke University Medical School Library

European Organization for Rare Diseases (EURORDIS)

Amy Farber, PhD - Founder, LAM Treatment Alliance

Gate39 Media - Chicago, IL

Tricia Geary - Barrington, IL

Genetic Alliance

Beverly Goldfarb - Short Hills, NJ

Herb Goldfarb, MD - Short Hills, NJ

Scott Goldfarb - Barrington, IL

Shereene Grant - FAU, Boca Raton, FL

International Society for the Study of Vascular Anomalies (ISSVA)

Marnie & Jeff Kaufman - Adenoid Cystic Carcinoma Research Foundation

Keyplate Graphics - Fort Meyers, FL

Beth Mantey - Green Bay, WI

Cy Matheson - Durham, NC

Frank X. McCormack, MD - Scientific Director, LAM Foundation

Art Mellor - Founder & President, Accelerated Cure Project

National Institute of Health (NIH) Office of Rare Diseases (ORD)

National Organization for Rare Disorders (NORD)

National Organization of Vascular Anomalies (NOVA)

Annette & Robert Petersen - Co-Founders, Gorham.dk

Rare Bone Disease Patient Network (RBDPN)

Stanley G. Rockson, MD - Scientific Director, LRF

Stephanie Shore, - Dallas, TX

Erin Spera, RN, MS, CPNP - Children's Hospital Boston

Mary Beth Sylvia, MS, CFNP - Children's Hospital Boston

United States Bone & Joint Decade (USB&JD)