

# HELPING HANDS

FOR FRIENDS, FAMILY, AND SUPPORTERS OF THE CHILDREN'S GAUCHER RESEARCH FUND



THE ONLY THING INCURABLE IS OUR PASSION.



Valerie Yannias, Victoria Casares, Gregory Austin Macres, Noah Jerome Shaffer, Kyle Herrel, Jared Patrick Ashley, Joseph DeFacci, Andrew Dennis Doran Jr., James Tyler Cooper, Lauren Marsh, Ryan James Conklin, Cameron Robert Watson, Kristina C., Madeline Collin, Grant & Garrett Geyer, Hannah Colwell, Emma C. Pozzobon



*Greg Macres*  
**Gregory Macres**  
Chairman/Founder



*Deborah Macres*  
**Deborah Macres R.N.**  
Founder

Each of you who support the CGRF are making an enormous contribution to medical science. There is no doubt that one day the consequence of your compassion will be felt by parents and children around the world. The progress that has been made became abundantly clear at the Lysosomal Diseases and the Brain conference that was held in June 2006. Over 100

medical scientists from ten different countries assembled to discuss research that has never been discussed before. A few short years ago assembling this group of world-renown scientists, much less the science that was discussed, would have seemed impossible. We may still be in the beginning stages of finding a cure, but the momentum is clearly building. You are winning "the

*“To laugh often and much; to win the respect of intelligent people and the affection of children; ... to leave the world a little better; whether by a healthy child, a garden patch or a redeemed social condition; to know even one life has breathed easier because you have lived. This is the meaning of success.”*  
– Ralph Waldo Emerson

respect of intelligent people", and you are winning "the affection of children". Read Tanya's words on page 2 of this newsletter; a mother who is intimately involved, and a daughter who is "Wishing Upon a Star". Your collective efforts will undoubtedly leave the world a better place. "This is the meaning of success."

## The CGRF Funds 3 Years of Research

In 2003 and 2004, with your hard-earned donations, the CGRF funded two years of research with Dr. Tony Futerman at the Weizmann Institute of Science in Israel. The research focused on determining the mechanistic relationship between defective Ca<sup>2+</sup>-homeostasis via the ryanodine receptor (RyaR) and the type of Gaucher disease, i.e. types 1, 2 and 3.

Layman's Translation:

1. On a molecular level, how does the disease affect the neurons in the brain?
2. Why is the brain affected in type 2 & 3 Gaucher disease, but not affected in type 1 Gaucher disease?
3. Does the regulation of calcium, which naturally occurs within neurons, become defective, thus impairing the function of neurons in the brain?

The results of this research were published in the scientific literature in early 2005. Subsequent to this publication seven other relevant papers that relate to LSDs and/or defective calcium have been pub-

lished by a collaboration of Dr. Futerman's laboratory and others. Of great interest is the fact that defective calcium-homeostasis has now been reported in a number of lysosomal storage diseases, including Gaucher type 2, Niemann-Pick A, Sandhoff disease, and the GM1 gangliosidosis, and in a number of other lysosomal storage diseases

*“... the mechanisms first described in type 2 Gaucher disease (research funded by your donations) may be of relevance for understanding other LSDs, and possibly other neurological diseases.”*

(LSDs), implying that the mechanisms first described in type 2 Gaucher disease may be of relevance for understanding other LSDs, and possibly other neurological diseases.

The CGRF has recently funded a 3-Year Research Grant with the Weizmann Institute of Science to study up-and-down-stream pathways that may be altered in neuronopathic forms of Gaucher disease.

# Madeline Collin

# Wish

Upon A

# Star

My story begins twelve years ago. On Thursday 22nd September, 1994, I gave birth to my daughter Madeline. She was a healthy baby at 8-lbs. 13.5 oz. Family life carried on until early January, 1996, when Madeline was taken ill and diagnosed with Type 3 Gaucher Disease. Since that day almost eleven years ago my life has never been the same. Nothing prepared me for what the future had installed. At times I feel sad, frightened, angry and alone. The pain inside is sometimes unbearable. Coping is sometimes hard and I often ask why this has happened, what has my daughter done to deserve this?

Despite everything Maddie has been through she is a strong independent person and is eager to grow up and enjoy life. Every parent is proud of their child; I am no exception. As I write this article I want to sing from the treetops and tell you what an inspiration she has been to me and how proud she makes me feel.

Despite the ups and downs of living with a long term progressive illness Maddie's determination has given her the strength to cope with the many challenges that life has thrown at her. School and her lack of confidence in her abilities have been the main issues of concern. School has been

a major difficulty for Maddie. Her eye movement, short term memory and auditory processing problems associated with her condition have meant that she finds school very challenging. At times this has been exhausting and very stressful. But with a lot of patience, time, and effort from Maddie's teacher combined with Maddie's determination she has made it through primary school and sat her end of year exams. School will always be a challenge but she has a full-time support worker at school now and this will enable Maddie to reach her full potential by giving her the support she needs.

At the beginning of September, 2006 with my stomach churning and feeling slightly sick I said goodbye to my eleven-year-old daughter Maddie as she went off to secondary school for her first day. I spent the whole day worrying; every

hoping that this would boost her confidence. Maddie swims like a fish, plays the violin, dances each week and has recently started horse riding. When I see Maddie on a horse I can see that she truly loves to ride. She has this huge smile on her face, her back is straight and she oozes self confidence. It is the same with swimming. Maddie is a truly bright girl, and in the right environment with the right support and encouragement the world is her oyster. Take away the stress of everyday life, the pressure of time and the need to be good at everything and Maddie and many other children like her with similar problems would thrive.

Every now and then Maddie asks me questions that are hard to answer. These questions are mainly about what will happen to her in the future, and if there will ever be a cure. These questions



"Since that day almost eleven years ago my life has never been the same ... At times I feel sad, frightened, angry and alone ... often I ask why this has happened, what has my daughter done to deserve this?"

time the telephone rang I thought it would be the school saying there was a problem. Later that day at 3:30 pm she arrived home exhausted but with a smile on her face.

Because of the difficulties Maddie has had in school we have tried to encourage her to take up activities and interests outside of school and academia

always come out of the blue and turn my insides up side down. As a parent you always want to be honest with your children and I say "who knows what the future will bring". It is hard though to be positive when after 11 years we are really no further forward with a treatment for the neurological involvement of the disease.



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Maddie is currently on the Zavesca trial for Type 3 Gauchers disease in combination with enzyme replacement therapy. She has been in the trial for over three years now and as the trial comes to an end we all wait in anticipation for the results. She is scared that there will never be a cure and as she gets older she has begun to resent all of the medical intervention. Whenever Maddie and I are together and we see a shooting star we look at each other and know that each other have made the same wish - A wish for a cure.

A year or so after Maddie had been diagnosed with Gaucher Disease Susan Lewis of the UK Gauchers Association asked me to join the executive committee as the representative for neuronopathic families. I had reservations. I thought what do I know about charity work and I was just a mother struggling to come to terms with Maddie's illness. I think it was the stubborn streak in me that saw it as a challenge. Also I thought back to when Maddie had been diagnosed and remember how alone I had felt and I thought if I could just help another family and let them know that they were not alone, then it would be worth it. Two cases I remember clearly are when we traveled to Nottingham to meet a family whose daughter had just been diagnosed with Type 3 Gauchers disease. I took Maddie with me and when they saw her and saw how normal she was and that she was able to do lots of things I could see that they saw hope. Another case was just before Christmas one year and Dr. Vellodi,

Maddie's Consultant at Great Ormond Street Hospital, asked me if the mother and father of a type 1 child could come up to the ward to speak to me. When they arrived they asked where Maddie was, at that moment Maddie rushed past the ward door totally naked with tinsel in her hair; again I saw hope.

In the nine years since I became involved with the Gauchers Association I have tried to offer support to patients and their families, raise the profile of neuronopathic Gauchers disease, improve the clinical management of patients and understand the challenges they face when accessing education. Most if not all of this has been achieved in partnership with Dr. Ashok Vellodi.

Looking back I am particularly proud of three things that I have achieved. I am just a parent who wants to be able to give my daughter a chance at life.

- Hosting two Neuronopathic Family Conferences in 1999 and 2004 for UK and European families.
- The development of the European Consensus on the Management of NGD and its publication in the Journal of Inherit Diseases in 2001.
- Being a contributing author in the new book on “Gauchers disease”.

Not long after I got involved in the Gauchers Association I received an e-mail from Greg Macres, the father of Gregory Macres who sadly passed away at the age of four-years-old from complications after a bone

marrow transplant. Several e-mails and a long late night telephone call across the Atlantic Ocean later, Greg had shared he and his wife Deborah's plans for the 'Children's Gaucher Research Fund'. Greg and Deborah's determination, energy and commitment to find a cure for neuronopathic Gaucher disease is a true success story. Understanding what happens in the brain of these children is vital and due to the rarity of the disease it is difficult to access money for vital research. The Children's Gaucher Research Fund (CGRF) has enabled professionals interested in the field of Gauchers disease to apply for funds to try and understand the disease with the ultimate goal of finding a cure.

In 2004 I was lucky enough to attend the 2004 Lysosomal Diseases and the Brain conference in Washington D.C. where I met Greg and Deborah Macres for the first time.



“Whenever Maddie and I are together and we see a shooting star we look at each other and know that each other have made the same wish - A wish for a cure.”

Many of the key Professionals in the field of Lysosomal Diseases from all over the world were at the conference. Greg and Deborah's vision to bring the lysosomal disease research community together was an important one and at this time it became apparent that advancement in one disease could potentially benefit other diseases. Following the success of the 2004 conference, in 2006 another Lysosomal Diseases and the Brain Conference took place in Sacramento, California.

In 2005 the Executive Director of the UK Gauchers Association retired and the Trustees of the Association asked me to take over the role as Executive Director. In this new role I now manage the day to day operational aspects of the Association including organizing conferences; publishing a half yearly newsletter; supporting families; liaising with Pharmaceutical and Homecare Companies and encouraging Research in Gaucher disease. I am also the convenor of the European Gauchers Association (EGA) which is made up of 24 European Gaucher Patient Organizations. The role of the EGA is to exchange relevant information in particular on: A) The treatment and management of Gaucher disease and research being carried out in member countries and elsewhere B) To keep member organizations aware of developments in member countries C) To encourage multinational research into Gaucher disease.

Taking on this role has certainly been a challenge but one I have enjoyed, one I thrive on, and one I feel extremely lucky to carry out.

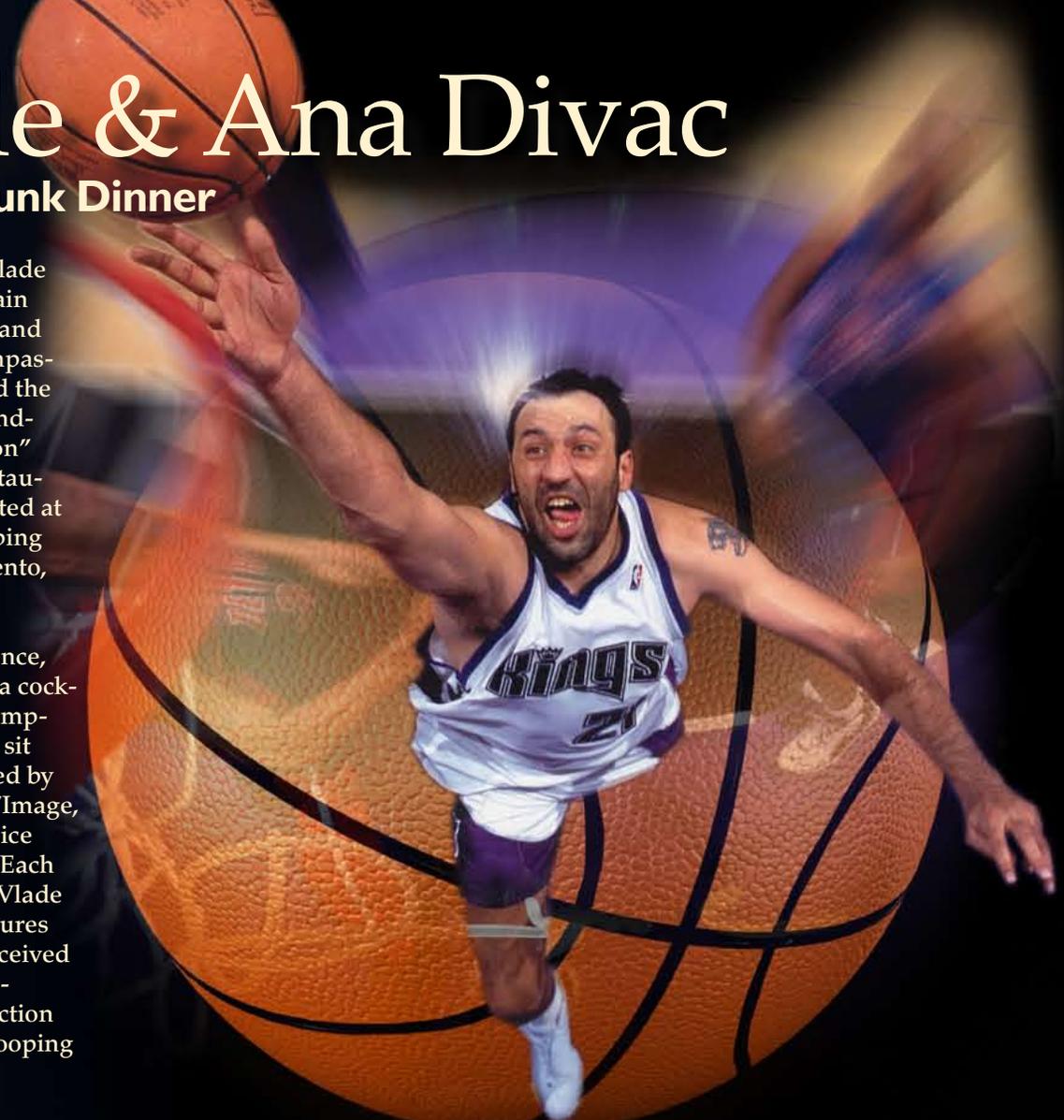
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# Vlade & Ana Divac

## A Slam Dunk Dinner

On July 24, 2006, Vlade and Ana Divac again offered their time and their words of compassion as they hosted the second annual "Find-A-Cure Celebration" dinner at their restaurant, L'Image, located at the Pavilion Shopping Center in Sacramento, California.

With 48 in attendance, everyone enjoyed a cocktail hour with scrumptious appetizers, a sit down dinner served by the fine chefs at L'Image, and a glorious choice of desert specials. Each guest visited with Vlade and Ana, took pictures with Vlade, and received a framed and auto-graphed 8" X 10" action photo of Vlade swooping to the basket.



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# 10<sup>th</sup> Annual

## Gregory Austin Macres Memorial Golf Tournament

The Gregory Austin Macres Memorial Golf Tournament began in 1997 shortly after little Gregory Macres passed away during a bone marrow transplant. Over the past 10 years the sponsors and volunteers who work countless hours for this exceptional event, have been instrumental in raising substantial funds for medical research.

September 28, 2006 marked the 10th Annual Gregory Austin Macres Memorial Golf Tournament benefiting the Children's Gaucher Research Fund. Held at Cinnabar Hills Golf Club in San Jose, California and sponsored by Coldwell Banker Silicon Valley, the full-field shotgun tournament was enjoyed by over 150 golfers. We thank the tee sponsors that included local title, mortgage and pest control companies. After 18 holes of golf, the golfers enjoyed a cocktail hour, a silent auction, dinner, and raffle awards.

John Carman's Bike Brigade - "Four years ago an idea came to me; let's raise awareness and raise funds for the cure of

Gaucher disease. Not only would it touch the lives of those affected by Gaucher disease, but the research may benefit other diseases. I decided to ride my bike to the Gregory Austin Macres Memorial Golf Tournament - approximately 45 miles. Would anybody sponsor the effort? Yes indeed! Compassionate individuals donated their time and their money raising several thousand dollars. In the 2nd, 3rd and 4th year we increased the participation and donations through the cycling efforts of many others who joined this effort."



# Suzy Ashley Helps Raise Funds

The Children's Gaucher Research Fund has earned over \$16,000 so far and you can help us earn more by simply switching stores!

We became involved with a company that is providing us with environmentally safe cleaning products for use in our homes. These are unique and high-quality natural products at reasonable prices. From everyday products like soap and shampoo to vitamin and mineral supplements - each product is scientifically designed to improve the well-being of you and your family.

We have several board members using these wonderful products and they love the results. These products will help everyone keep a cleaner and safer atmosphere in their homes. If you are

concerned about your overall health and wellness, this company can help you. They have also provided us with a unique opportunity to participate in a residual fundraiser that will help fund our research. To date we have earned over \$16,000 by participating. If you would like to learn more about these products, or about how you can participate in this fundraising opportunity, please call or email.

Sincerely,  
Suzy Ashley  
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## 100% To Research

You need to know:

1. The CCRF is a **legitimate** IRS approved 501 c3 non-profit organization.
2. **100% of every donation** goes to medical research.
3. We **do not** hire professional fundraising companies who keep 50% of donated funds.
4. We have talented volunteers who **donate** their time and talent for a variety of our needs.
5. All administrative costs are paid for by the **founders**.

Simply put:

If you send your hard earned dollars - **It ALL goes to medical research.**

Visit our web site at:  
[www.childrengaucher.org](http://www.childrengaucher.org)  
All family stories can be  
read on the website.

**Contributions Payable To:**  
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## Children's Gaucher Research Fund



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