

# HELPING HANDS

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



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 Danielle Loncharich Emma C. Pozzobon

THE ONLY THING INCURABLE IS OUR PASSION

# BABY AALIYAH



We are writing to share with you a commitment we have made. We have joined hands with families from across America. Our Goal: to find a cure for the disease that took our daughter Aaliyah.

We lost our daughter Aaliyah to Gaucher disease type II on June 26, 2008, which was a big topic here in Grand Rapids and in

various places around the world, primarily in the Gaucher community. Aaliyah is very well known in the "medical mile" for her frequent hospital visits/admissions. She also has been

in the news numerous times, she has been featured in the newspapers and also on NBC's Today Show as well as Dateline would like to feature Aaliyah's story (My husband is just not ready yet; the pain of losing her is still too fresh). It is impossible to describe to you the pain of losing our child. Aaliyah's short life changed our lives and so many others lives forever. Now, after losing Aaliyah, we have choices to make. We can give up on life, or we can help give life to other children who suffer from this terrible disease. We know that Aaliyah is cheering us on. We know that Aaliyah is encouraging us to be positive, to be proactive, and most of all, to help make a difference. We know that Aaliyah is encouraging us, and we believe guiding us to find a cure once and for all!

**"We can give up on life, or we can help give life to other children who suffer from this terrible disease"**

You may be thinking, "Oh no, I'm on another mailing list"! Quite frankly, you are, but remember, YOU can help make a very big difference in the lives of children like Aaliyah with this life threatening and debilitating disease. There is some apprehension in writing this article. However, after joining hands with these other families, writing this article becomes easier. We think

of the young woman whose 20-month-old daughter was recently diagnosed with Gaucher ('go-shay') disease. As she shares the story of her daughter's endless

medical needs, we are reminded of our own lives, not too long ago. She then breaks down in tears, and while sobbing and distressed, asks, "Am I going to watch my baby die before my eyes"? To tell you these are painful reminders is an understatement. But it does underscore two facts:

1. This may not be our tragedy. But it's today's tragedy for many families.
2. We know all too well that the answer to her question may be, yes.

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



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

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In short, this is the type of real life story that allows us to shelve any embarrassment and ask you to become involved in a grass roots effort with this research fund. We don't wish to ask for a large commitment of time, simply a decision that your charity of choice, will be the "Children's Gaucher Research Fund", which is 100% tax deductible. From that point forward this may entail sending a periodic donation, or simply developing an awareness of opportunities that may help raise research funds.

Here is what initially caught our attention and solicited our respect:

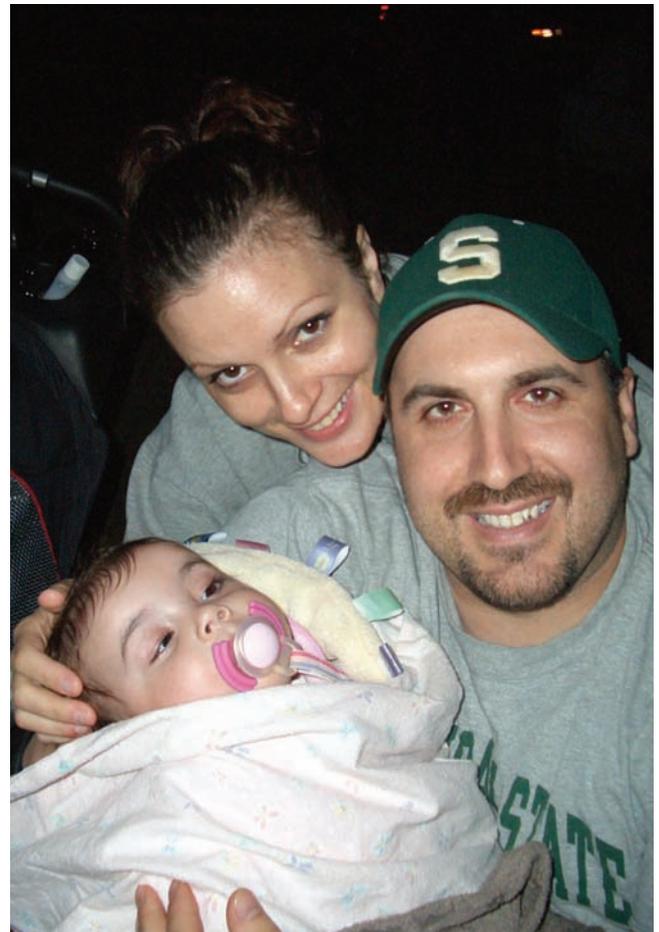
1. 100% of donations go directly to medical research. There are no salaries. There are no administrative costs. All costs for postage, printing, web site development, etc. are either donated, or paid for by the founders.
2. This is a grass roots effort, whereby parents across the nation, and beyond, are working together to raise funds for medical research. We as parents are not asking for a handout, we are asking for your "Helping Hand".
3. These are children, who only ask to have a chance at life. As a group of parents, we are willing to fight the battle with and for our children, and we have decided to be on the front lines of the battlefield.
4. It has been said that the 21st century is the century of the brain. This research will have overlapping benefits; benefiting a host of adult and childhood diseases, that affect the brain.

In one of the past Helping Hands newsletters this quote seemed to crystallize our feelings:

"Over the years of my life, I have sporadically donated time and money to various causes. I never really found a charity I could adopt, a charity I could believe in, one that captured my heart. We have all heard stories about charitable causes where only a small percentage of donated funds are applied to the charity itself. In the past, this has given me cause for concern and has made me cautious. I was never able to find a charity that I could call, my "Charity of Choice". I have been taken by your efforts and your commitment. I am proud to choose the Children's Gaucher Research Fund as my charity of choice."

We understand that you may already have a charity that you sponsor. However, if you feel a personal connection, as we do, we ask that you consider making the Children's Gaucher Research Fund your "charity of choice". Our family will be a part of the Children's Gaucher Research Fund, until a cure is found, and it is people like you that WILL make that possible!

**TIMOTHY & ELIZABETH HEINZELMAN**  
Kentwood, MI (carepage name is: BabyAaliyah)





## High School KEY Club Students

“RAISE A RECORD \$21,000”.

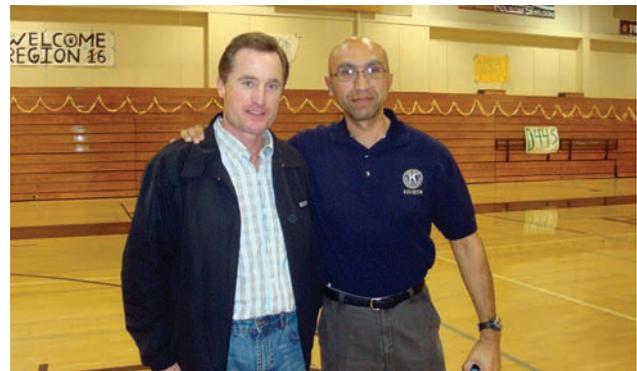
What is KEY Club? This is a question I am always asked while partaking in a community service event where my students are volunteering. Most people in the Sacramento area and vicinity have little or no idea what the “K-E-Y” in KEY Club stands for. Kiwanis Educating Youth (KEY) is a part of Kiwanis International, an organization striving to educate young men and women to volunteer spare time in their local communities. This organization develops and mentors youth as leaders, builds playgrounds, raises awareness or funds for pediatric research, and many more local projects. No problem is ever too big or too small!

For the last four years, I’ve had the immense pleasure of being a faculty advisor and mentor to a magnificent number of extraordinary young men and women. I sometimes can’t believe (and often forget) what teenagers are capable of doing—when given proper guidance and instruction.

.....  
*“... a student led organization raised more than \$21,000 for a single non-profit organization or charity. The energy of that one event was electrifying!”*  
 .....

As I mentioned above, four years ago I too had no idea what KEY Club was all about. I begrudgingly agreed to become their advisor—after a colleague of mine transferred schools—not knowing what I was getting myself into. I muddled my way through the first events and was completely shocked when I attended my first Florin High School Awake-a-thon. The purpose of the event was to raise funds and awareness for the Children’s Gaucher Research Fund, while staying awake for an overnight stay in the school’s gymnasium and multipurpose room. I anticipated one hundred students, maybe two hundred at the most, not the close to three hundred that actually attended that first year. Our endeavors rose close to \$8,000 and provided the club with the determination to raise even more money the following year.

Previous awake-a-thons had taken place at our school site, but not as significant and recognized as what the Florin High School awake-a-



thon would become. For the last three years we’ve welcomed secondary school students throughout the Elk Grove, Sacramento, and Davis Unified School Districts. Every year since, our numbers continue to grow. And, last November more than 600 students joined in the effort to find a cure for Children’s Gaucher Disease. With the increased number of participants, the amount of pledges raised also increased significantly! For the first time in the twenty-year history of our school, a student led organization raised more than \$21,000 for a single non-profit organization or charity. The energy of that one event was electrifying!

Although the academic year is coming to an end, the student leadership of KEY Club continues to look ahead to the fall for the preparation of this yearly event. I enjoy seeing the positive attitude and vitality that this event brings out of these young adults. Their determination, aspirations, and hopes are contagious. Imagine what could be accomplished if every school had a similar student organization?

**ELIAS MENDOZA**  
 Teacher / Key Club Advisor  
 Florin High School  
 Sacramento, California

## A Gaucher Mouse

RECENT RESEARCH FUNDED BY THE CGRF.

There is currently not effective treatment for the neurological problems of patients with neuronopathic Gaucher disease (also called type 2 or acute neuronopathic and type 3 Gaucher disease or chronic neuronopathic). In order to develop such treatments there is a need to better understand the disease and to test the safety and efficacy of new therapies before they are tried in patients. Therefore, a mouse model for the disease is very important. Currently, there are a few mouse models for the disease but they all have significant limitations.

Years ago a mouse model was made with a complete inactivation of the Gaucher gene – a “knockout mouse”. Unfortunately such a mouse is not viable and most of the pups that are born die within one day. Other mouse mutants with specific human mutations were subsequently produced (“knock in mice”) but they were either completely normal, especially in the brain, or they were not viable just like the original knockout mouse. Last year the group of Dr. Stefan Karlsson from Lund, Sweden produced the first real neuronopathic Gaucher mouse. They managed to completely inactivate the Gaucher gene in the brain but the gene and therefore the enzyme (glucocerebrosidase) remained normal in the skin and allowed the pups to be born. However, the mice of this model all die by age 3 weeks. Although the ‘Karlsson mouse’ is very useful to study many aspects of the disease, it has a very severe disease with a short lifespan, a little like a severe type 2 Gaucher patient. Therefore, this mouse does not reproduce the human disease faithfully since it has no glucocerebrosidase activity at all while all patients with any type of Gaucher disease have some residual enzyme activity.

“Thus, there is a need for a mouse that will live longer and will reproduce better the human disease.”

Thus, there is a need for a mouse that will live longer and will reproduce better the human disease. This mouse has to live long enough to allow the testing of novel therapies. In order to accomplish that, the CGRF funded Dr. Avraham Yaron, Professor Tony Futerman and Dr. Yael Pewzner-Jung from the Weizmann Institute of Science in Israel to make a special mouse. This new Gaucher mouse will be a conditional inducible mouse.

The enzyme level in the brain cells of this mouse will depend on the amount of an antibiotic called tetracycline that will be present in the water that the mouse will drink. The higher the dose of tetracycline that the researchers will give this mouse, the lower the enzyme level will be. This new system will allow the researcher to lower the enzyme activity in the brain at will and at any age. Stopping the administration of tetracycline will lead it to recover of the amount of enzyme in the brain of the mouse. This recovery will be a way to test the extent in which replenishing the missing enzyme will reverse or prevent the neurological abnormalities in the mouse and will probably also be relevant to the treatment of the human patient. Therefore, if all goes well technically this will be a very exciting mouse that will be like Gaucher type 2 (lots of tetracycline) or like Gaucher type 3 (a little less tetracycline). We hope that this animal model will be ready in about 18 months.



**RAPHAEL SCHIFFMANN M.D.**  
Director, Institute of Metabolic Disease  
Baylor Research Institute  
Dallas, Texas

## Parkinson's disease - a window to neuronopathic Gaucher?

I am an Assistant Professor of Neuroscience at The Parkinson's Institute in Sunnyvale, CA. As my place of work suggests, my background is in Parkinson's disease (PD) and related neurodegenerative disorders, typically associated with aging. Some intriguing genetic and neuropathological studies indicated that an association exists between PD and type I Gaucher disease, and consequently, lead to my attending the Lysosomal Diseases and the Brain conference in 2006. This meeting was an eye-opening experience for me. It was filled with leaders in the Gaucher field, and properly sized and organized to promote interactions between scientists. My discussions from that meeting fostered the formulation of new ideas and hypotheses to test. I subsequently began pursuing the biological, mechanistic basis for the link between Gaucher disease and PD. My studies have revealed that there are enhanced levels of specific protein involved in PD, alpha-synuclein, in a model of Gaucher disease. These observations suggest that interactions between this protein and lipid components may contribute to the vulnerability of brain cells in Gaucher disease.

“...this meeting provided a forum for cross-fertilization between fields (Gaucher and PD)...”

I had the opportunity to share and discuss these findings, and their potential relevance, with the leading Gaucher scientists at the 2008 Lysosomal Diseases and the Brain conference. This conference provides an open environment for the exchange, development and challenging of new ideas between scientists. I cannot stress enough the importance of this sort of interaction in scientific advancement. In particular, this meeting provided a forum for cross-fertilization between fields (Gaucher and PD), and for me, has lead to potentially fruitful collaborations and a new research direction. Indeed, such collaborative efforts between fields could provide a new window of research into lipidoses, as well as PD, and may provide new targets for therapies for both. I am sincerely grateful to the Children's Gaucher Research Fund for their sponsorship of this forum.



**AMY MANNING-BOG**  
The Parkinson's Institute  
Sunnyvale, California

### Seeking Pro-Bono Communication Professionals... for a New CGRF Website

After ten years it is time for the CGRF to build a new website to incorporate the latest in internet technology – a task that is long overdue. As you know, we are able to commit that 100% of all donations go toward medical research because so many talented volunteers have offered their services – allowing us to minimize administrative and development expenses. We are launching an online development initiative and we are seeking pro-bono services from the following communications professionals:

- writer
- graphic designer
- video designer
- online marketer / marcomm manager

Ideal candidates will have strong portfolios showing expertise in one or more of the following:

- cause marketing
- social media
- medical, health, and/or wellness industries
- children and/or family services

If interested please email us at [research@childrengaucher.org](mailto:research@childrengaucher.org)

STANTON & KRISTEN ENOMOTO

555 HAHAIONE STREET, APT. 4G

HONOLULU, HAWAII 96825

ENOMOTO.OHANA@GMAIL.COM

February 11, 2009

Mr. & Mrs. Gregory Macres  
Children's Gaucher Research Fund  
P.O. Box 2123  
Granite Bay, California 95746-2123

Dear Mr. & Mrs. Macres:

**Subject:** Donation on behalf of Grace and Elsa Enomoto

We are writing to you to express our gratitude for your work on researching Type 2 and 3 Gaucher's Disease and support for families with children who are suffering from this disorder. We ask that you indulge us for a moment as we would like to share the story of our wonderful daughters and how they have brought us in touch with your work.

We are the proud parents of four beautiful daughters: Cameron, Skye, Elsa and Grace. Cameron is five and in kindergarten, and Skye is three and in pre-school. Sadly, our two youngest girls, Elsa and Grace, both passed when they were just a few months of age.

Elsa was born on January 23, 2007 and unbeknown to us at the time, displayed some indications of Gaucher's Disease such as stiffness, ichthyosis, laryngomalacia, and slow growth. We believed that she would out grow these conditions and, with the exception of her breathing difficulties, appeared to be doing so. Not knowing any better we assumed that she was developing normally. However, on April 27, 2007, she stopped breathing during her morning nap and did not recover. We were devastated. The autopsy findings listed the cause of death as undetermined. As you might expect, we were left with many questions that only compounded our grief in losing our 3-month old baby girl.

With the support of friends and family, we slowly moved forward with each day being a little brighter and better able to keep life in perspective. Through this, our fourth daughter, Grace was born on April 23, 2008. Like her sister Elsa, she unfortunately displayed similar symptoms and it prompted much closer observation and testing. When she was three months old, Grace was treated for her laryngomalacia and later underwent a fundoplication to address recurring bouts of reflux. She also underwent a series of tests for metabolic disorders and in August 2008 we received the heart-breaking news that she (and most likely, Elsa) had Type 2 Gaucher's Disease. After receiving this diagnosis, Grace remained in-patient at the Kaiser-Permanente Hospital here in Honolulu. Additional testing was performed to identify exactly where the mutations occurred in Grace's genetic structure and the results were entered into the Genzyme Corporation's Gaucher's Registry.

We were extremely fortunate to have a dedicated team of physicians, palliative care specialists, nurses, social workers, and counselors to help us not only understand the etiology of Type 2 Gaucher's Disease, but more importantly, they provided all possible comfort and support to Grace and helped our family prepare for the difficult months that lay ahead.

In Hawaii, none of the medical professionals (some with over 30 years of experience) could recall encountering a case of Type 2 Gaucher's Disease, and consequently, we were all learning side-by-side. In this regard, websites such as yours, provided us tremendous insight into Gaucher's Disease, an understanding of what to expect, and comfort in reading the stories of other families and knowing that we weren't alone.

Throughout the fall of 2008, the neurological and physiological impacts of Gaucher's became incrementally more pronounced in Grace. Thankfully, because we continued to be in-patient, we were able to receive appropriate care and comfort medication whenever it was necessary.

In the evening of November 18, 2008, our baby Grace passed away peacefully in our arms and was surrounded by her sisters, grandparents, and an incredible group of hospital staff who loved her as their own. Grace was almost seven months old. A small memorial service was held and it is through the generosity and love of our family and friends that we are able to make this contribution on behalf of both Grace and Elsa.

Please accept the enclosed donation and we request that it be used towards research on Type 2 and/or Type 3 Gaucher's Disease in the hope that it will expand the body of medical knowledge, provide better comfort and care to children like Elsa and Grace, and ultimately, lead to a cure for this rare disorder. We also ask that you provide us with a letter confirming receipt and acceptance, and that you keep us informed on the progress of the research your organization is funding.

With warm regards,

Stanton K. Enomoto  
Kristen K.H.C. Enomoto



Enclosure: Check #136, Payable to Children's Gaucher Research Fund



## 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:

Children's Gaucher Research Fund

P.O. Box 2123

Granite Bay, California 95746-2123

IN LOVING MEMORY OF

## JOSEPHINE ROSE LAMPITT

*"Josephine -- our precious gift who reminded many in her short life of the message of faith, hope, and love. We will spend our lifetime loving you until we are reunited together again in Heaven."*

APRIL 20, 2008 TO FEBRUARY 16, 2009  
WILLIAMSBURG, VIRGINIA

## Children's Gaucher Research Fund



8110 Warren Court  
Granite Bay, CA 95746 USA  
916 797 3700 tel  
916 797 3707 fax  
[research@childrengaucher.org](mailto:research@childrengaucher.org)

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