

Summer 2013

HELPING HANDS

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



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Grant & Garrett Geyer

Danielle Loncharich

Gregory Austin Macres

Three Legs of the Stool

By Greg and Deborah Macres

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- #2 ANIMAL MODEL
- #3 FDA/EMA APPROVAL

In each newsletter we try to give an update as to the progress that has been made in our effort to find a cure. In this issue, we decided it would be important to outline the “method to the madness” – meaning, is there a strategy to the efforts of the CGRF. To date, thanks to your generosity we have raised over \$2 million. That is a lot of money that has been given by thousands of hard working people who believe in and support our effort. But the truth is, in the grand scheme of medical research – in the grand scheme of finding a cure for these childhood diseases – it often takes significantly more than \$2 million. Thus, we need to ask the question, “What is our strategy?” How do we intelligently and efficiently appropriate dollars for maximum result? How does the CGRF leverage each and every dollar we allocate to research, so that the result has a multiplied impact on research, and an eventual cure?

We are mindful of the fact that often, rare diseases do not attract interest or funding and sit idle for many years with little progress. Why does this happen? Our conclusion is that without the building blocks in place, it is more difficult for research laboratories to devote resources to pursue a particular disease. The CGRF is laying the foundation with these building blocks – which we refer to as the “Three Legs of the Stool”. By doing this we are giving research scientists around the world a running start – their path is easier to pursue – their path is more compelling – they have a better chance at success – we are fostering interest in, and funding for neuronopathic Gaucher Disease as well as 26 other Lysosomal diseases that affect the brains of children. So what are the “Three Legs of the Stool”?

#1 BASIC SCIENTIFIC RESEARCH

Have the initial Building Blocks of Understanding the disease been established through basic scientific research? If not, it is difficult for a scientist to feel confident that their thesis is valid. Since 2001 the CGRF has

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Deborah Macres
DEBORAH MACRES R.N.
Founder



Greg Macres
GREGORY MACRES
Chairman/Founder



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funded basic scientific research in the laboratory of Dr. Tony Futerman at the Weizmann Institute of Science in Israel. This research has answered many of the basic questions – affected locations in the brain – how brain neurons are impaired – the process of inflammation – etc. The outcome of this research has resulted in 12 different publications in various scientific journals.

“The CGRF is laying the foundation with these building blocks – which we refer to as the “Three Legs of the Stool”

#2 ANIMAL MODEL

Is there a viable animal model that can be used in scientific studies? If not, scientists have to prove their thesis in a petri dish – a much more difficult task. In 2008 the CGRF funded the creation of an Inducible Neuronopathic Gaucher Mouse. This animal model is now successful, and in late 2013 will be available to any research laboratory in the world. Thus, research laboratories will now have a viable animal model to use in their research.

#3 FDA/EMA APPROVAL

Is there a roadmap for FDA (Federal Drug Administration) and EMA (European Medicine Agency) approval? If not, this is a task that can take years of preparation. We have now set our sights on the third leg of the stool - creating a Roadmap for FDA/EMA Approval. This requires the accumulation of both clinical data and molecular bio-markers that have been amassed by researchers in the US, Europe, and Asia. It is a herculean task to accumulate one central depository of data, and then find agreement among the scientists in identifying the appropriate bio-markers and clinical outcomes. This is required by the FDA and the EMA, and this “Roadmap” will become freely available to any research laboratory in the world.

Over the past 10+ years the CGRF has accomplished the first two legs of the stool. Once this third leg is complete, the CGRF will have successfully paved an easy road, motivating research laboratories to devote more time, resources and funding for these lethal childhood diseases. They will have the Basic Science – they will have an Animal Model – and when they discover a successful therapy they will have a path to FDA/EMA approval.

We believe this is the intelligent approach. We believe this approach leverages the impact of your hard earned dollars. And, we know in our hearts that this incredible progress would not have been possible without your support. For that, we humbly thank you.

GAUCHER (NEUROPATHIC) THERAPY INITIATIVE (GTI) ...The third leg of the stool – FDA/EMA Approval

By Raphael Schiffmann

On January 28-29, 2013, a meeting took place in Atlanta, GA on creating a ‘road map’ for the development of treatment for neuronopathic Gaucher disease. The meeting was organized by the CGRF (Mr. and Mrs. Macres) with the participation of experts in Gaucher disease from the US and abroad and of senior representatives from 4 biotechnology/pharmaceutical companies. These companies are already involved in therapies for rare diseases including Gaucher disease and they also provided unrestricted funds for the meeting. This important meeting is one of “3 Legs of the Stool” (see article on page 1) on which rests the progress towards effective therapy for neuronopathic Gaucher disease (nGD). The details of clinical trials for nGD will have to be defined once a specific medication is ready for testing in patients. However, it is very important to be prepared for future therapeutic studies by defining how nGD patients present to doctors and how the disease manifestations change over time (natural history). It is also important to be ready with specific tests for nGD because they will be the means by which the efficacy (safety and effectiveness) of a future test therapy will be determined (clinical outcome measures or endpoints).

The two-day January 2013 meeting started with summaries

of what we know about how the health problems of patients with type 2 and type 3 Gaucher present. An agreement was reached about the definition of nGD – in order to diagnose nGD the patient should have at least the eye movement abnormality typical in these patients (also called supranuclear gaze palsy by doctors in the field). Professor Tony Futerman from the Weizmann Institute of Science in Israel, whose work on nGD is funded by the CGRF, briefly summarized where we stand in terms of using animal models to understand nGD. Based on studies in animal models of nGD, Prof. Futerman’s laboratory is expected to inform the scientific community in the coming months

whether a future therapy that addresses the basic biochemical defect in nGD should be expected to reverse existing neurological manifestations or just to prevent the disease from getting worse. The structure of future clinical trials in nGD largely depends on the answer to that question.

A detailed discussion of the natural history of nGD followed and led to the structuring of possible future clinical trials as well as the description of likely clinical outcome measures. The meeting concluded with a plan to move forward with this “road map”. This plan includes summarizing the natural history of nGD based on data from centers around the world; developing and implementing a standardized neurological examination of nGD patients; an effort to discover biomarkers (chemical changes) in blood and spinal fluid of patients with nGD that will help with therapy evaluation; moving ahead with initial contact with the FDA (Federal Drug Administration) and EMA (European Medicines Agency). A follow up meeting in order to continue building and developing the GTI will be planned for late 2013. It is important to emphasize that this is a comprehensive and international

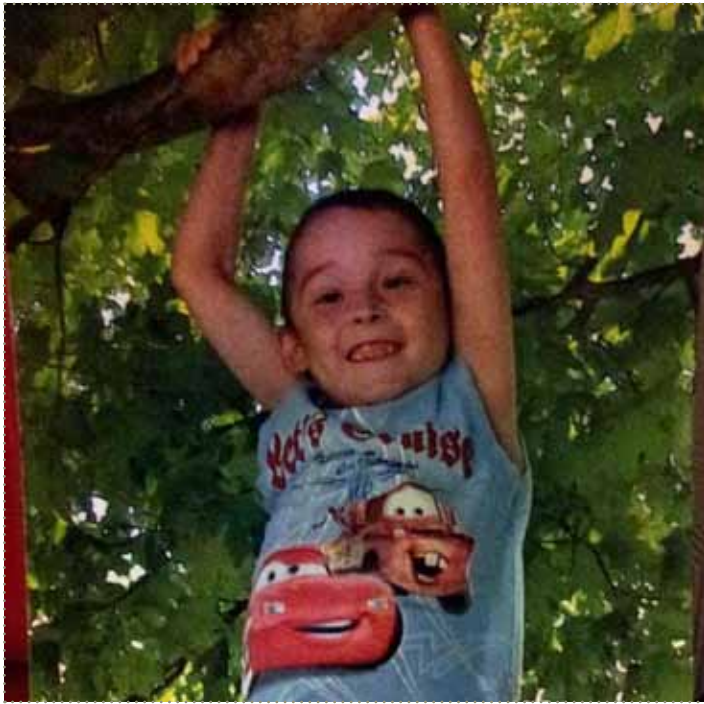
“I cannot emphasize enough how important each leg of the stool is in moving the field of medical science forward to the point that one day, there will be hope for these children and their families.”

effort that should include all nGD centers around the world that are interested in collaborating on an effort to facilitate and optimize future clinical trials in nGD.

I cannot emphasize enough how important each leg of the stool is in moving the field of medical science forward to the point that one day, there will be hope for these children and their families.



**RAPHAEL SCHIFFMANN
M.D., M.H.Sc.**
Baylor Research Institute
– Texas – Chairman, CGRF
Scientific Advisory Board



Brandon Ray Buckler

We lost Brandon Ray to Gaucher's Disease on May 1st, 2012. Brandon was twelve years old at the time of his death. Brandon's words on his death bed were very strong in the belief that he would beat his horrible disease. Brandon Ray dreamed of being a normal little boy, always hoping and praying that a cure was to be found in his near future. When being sent home from the hospital for the last time, with no hope of survival, only to succumb to this horrible disease, his family holding both of his hands while praying for a miracle. Brandon Ray's Deacon from his favorite nearby local church spent hours with Brandon and his family during this four days of this grueling, emotional roller-coaster. Our Deacon was having a conversation with Brandon about this point in his life's journey; Brandon Ray was listening, but not verbally responding all the time. He would squeeze your hand or blink his long lashes to let you know he could hear you and understood what you had said. In the middle of this one particular conversation, the Deacon asked Brandon, "When you meet God, give Jesus a high five and put in a good word for me." Brandon in his tired barely audible voice said, "I will not tell him anything (short breath taken) because (moment of silence and a deep breath, again) with a louder voice, I'm not going to see him!" You could hear Brandon struggling to breathe, he took an even deeper breath and spoke even louder than before, at this point the entire room of family and friends were listening. The whole room heard his next statement, **"I AM GOING TO FIGHT!"** He passed away within the next twenty-four hours, after speaking those words with so much faith, strength and belief in his voice.

Brandon was only a child, but throughout his life he was loved by many and has inspired them within their own lives to better themselves personally, strive for a better quality of life and helped them realize they can triumph in all of life's adventures. The pain of losing a child is indescribable. The pain of losing Brandon Ray from our world has inspired people who didn't even know him, to love him and use his example of pure strong determination of self will and his

love for life, as their daily inspiration. The loss that we and our family have suffered just reminds many of us, what is most important in our lives. Brandon Ray is a source of strength to us not only in life, but in death as well. His courage and attitude in the face of adversity is an inspiration to everyone who had the joy of knowing him. It is with his spirit that we want you to join us to spread the word of this Little

"Brandon Ray dreamed of being a normal little boy, always hoping and praying that a cure was to be found in his near future."

Inspiration and help us keep Brandon Ray's memory alive. Inspiring other children and families to reach beyond their restraints in life and keep fighting - hoping - and believing - we will find a cure. We know that Brandon Ray is encouraging us to find a cure and to share his story with the world.

In June of 2012 and again in June of 2013 our family and friends host a Brandon Ray Buckler Memorial Rodeo with a portion of the proceeds benefiting the Children's Gaucher Research Fund. Prior to his death, Brandon Ray loved the idea of his own Rodeo. He loved the fact that his rodeo would educate people about his disease and make more friends. Prior to June, 2012, he was excited as he would count down the days until the Rodeo; it just wasn't meant to be. He died the month before. After looking back, Brandon was counting down the days until he was to meet God. We decided after his funeral, to take a page out of Brandon's way of living life. Brandon Ray would have said, "The rodeo must go on!"

Brandon Ray's spirit was at his Rodeo. You could feel his presence from the beginning of the Rodeo's display of the United States Flag flying around on a barrel horse, to the releasing of the balloons for

“The whole room heard his next statement, I AM GOING TO FIGHT!” ”

our little angel in heaven. The announcer’s voice was breaking up as he told Brandon’s life story to the audience full of families and friends. Cowboys/Cowgirls quietly listened, with tears in their eyes, hats on their chest, and heartfelt pain on their faces. Two young bull riders rode their bulls with Brandon Ray’s initials on their vest. Neither bull rider knowing Brandon Ray personally, both putting BB over their hearts. One of the bull riders later said, “It felt like someone was on the back of the bull with him!” He stated, “normally I wouldn’t have stayed that long on that bull ... I have sat on the back of that bull a couple of times now, never made it past the first buck right out of the chute.”

At the end of the night we, along with the coordinator, her husband and the Rodeo owner’s wife sat under the stars discussing how wonderful the Rodeo turned out. An orange balloon fell out of the sky

and landed on the bench seat close to the coordinator and was gently, waving back and forth ever so slightly. We all looked at each other and went back to staring at the waving balloon. The Rodeo owner’s wife, Sandy, broke the moment of silence and said, “I think this is a sign from above, Brandon’s way of thanking you.” She walked away with tears in her eyes.

We very much appreciate your time, consideration and letting us share some of Brandon’s story. He was a wonderful child with a horrible disease.

RAY AND TAMMY BUCKLER
California City, Kentucky



Macres Heaven Birthday Ride 2013

April 13, 2013, marked the 16th heaven birthday of our precious son Gregory. We have chosen to honor his legacy through the Children’s Gaucher Research fund, and each year we celebrate his life with a 32 mile Memorial Ride visiting significant locations in his journey battling Gaucher disease. We thank those who supported the ride, and we are excited that this year we raised a record amount - \$7,700.

This year we again had a bit of excitement as we planned our journey to the Bay Area to celebrate Gregory’s Heaven Birthday. A few days before our ride our Golden Retriever was bitten by a Rattle Snake (nothing \$1,500 couldn’t fix – she is fine) but we did have to bring her with us, find a friend to watch her during the bike ride, and convince the Marriott to welcome her as a guest.

This year Deborah, Victoria and Nicole joined the 32 mile Heaven Birthday Ride. It was a beautiful day and it was definitely “memory lane” as we rode from the neighborhood I grew up in as a child – to the hospital that cared for Gregory – to the site of his memorial service – past the restaurant where we met for the first time – through our old neighborhood in Campbell – and on to Gregory’s resting place. Memories of our little boy are always with us, but especially on this special day – his Heaven Birthday.

Pursuing a cure holds great meaning for us, allowing us to build a positive legacy for our little boy. But most of all we are truly humbled; humbled by those who choose to support this journey, and humbled by your incredible generosity.





Play For Gaucher

In Honor of Lincoln Lewis Wheatley

On March 23, 2013, the “Play for Gaucher” Poker Tournament took place in Canton, Ohio. This annual event, now in its fourth year, has raised over \$11,000 for children’s Gaucher research. This event draws a crowd of roughly 60-100 players a year. This year’s event featured a raffle trip to Las Vegas, sports memorabilia, college football tickets, and much more! We have successfully taken something my team already likes to do (poker) and turned it into a larger event with a purpose. We strive to give participants a great value for their time and money. Although it is a charity event, we believe that if you follow that simple concept, that core principle...people will participate and the event will flourish naturally. It’s as simple as that!

With that being said, we have had our challenges. It has also been emotional and frustrating at times. However, our event has become easier to organize and manage year after year. We are able to learn from past experiences and have become much more efficient and effective at what works best. A team of 4 organizes this event, and each team member

takes on a separate responsibility which reduces the overall burden. We also leverage the power of email and the social networking (along with some old-fashioned word of mouth, as well). At this point, now that we have passed the initial first-and-second year anxiety and challenges, the event is fairly self-sustaining. Our team encourages anyone looking to organize such an event, for whatever the charity may be, to not give up and keep at it! Feel free to contact us at playforgaucher@gmail.com if you have questions regarding starting an event of any kind...we would be more than happy to offer tips and consultation on creating a website, using email, and general event promotion.

More details on the 2013 Play for Gaucher charity poker event can be found at www.playforgaucher.org. Don’t forget to register for next year’s tournament if you plan on being in the Ohio area.

ZACHARY AND KELLY WHEATLEY
Lewis City, Ohio

JESUS SALCEDO Compassion & Commitment



- Jesus understands our commitment that 100% of every dollar you donate goes to research.
- Jesus understands this requires that we keep expenses as low as possible.
- Jesus has become a part of this journey to help these children

For the past 8+ years Jesus Salcedo has offered his incredible talent to design these newsletters. I think you will agree he does a terrific job – and he offers all of this – for all these years – FREE OF CHARGE. We would like to recognize and

thank Jesus for “Working his Magic” and lending a “Helping Hand”. Jesus will hold a small piece of those future words from a physician to the anxious parents of a sick child – “Do not worry, we have a cure”.



ONLINE DONATIONS

can be made by visiting
www.childrensgaucher.org
OR

www.cgrf.org

All family stories can be read on the web site.

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.childrensgaucher.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

Children's Gaucher Research Fund

8110 Warren Court
Granite Bay, CA 95746 USA
tel 916 797 3700
fax 916 797 3707
research@childrensgaucher.org



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