#1 Basic Scientific Research

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”? 

#2 Animal Model

#3 FDA/EMA Approval

In this issue, we are presenting the “Three Legs of the Stool” to our friends, family, and supporters of the Children’s Gaucher Research Fund. The first leg of the stool is Basic Scientific Research. The second leg is the Animal Model. The third leg is FDA/EMA Approval. 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
On January 28-29, 2013, a meeting took place in Atlanta, GA on creating a ‘road map’ for the development of treatments 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 treatment 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 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.”

By Raphael Schiffmann

Gaucher (Neuropathic) Therapy Initiative (GTI) ...The third leg of the stool – FDA/EMA Approval

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 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 breath, 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 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...
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.
**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”.

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**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 experience, 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](http://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
In Honor of Lincoln Lewis Wheatley

60-100 players a year. This year's event featured a raffle trip for Gaucher research. This event draws a crowd of roughly 100-150 people.

On March 23, 2013, the “Play for Gaucher” Poker Tournament took place in Lewis City, Ohio. The event was hosted by a local high school and a group of friends. The two main goals of the event were to raise awareness for Gaucher disease and to raise funds for research.

We believe that if you follow that simple concept, that core team already likes to do (poker) and turned it into a larger network (along with some old-fashioned word of mouth, on-line donations, and much more!) We have successfully taken something my principle…people will participate and the event will flourish.

and much more! We have successfully taken something my principle…people will participate and the event will flourish. We also leverage the power of e-mail and the social networking (along with some old-fashioned word of mouth, on-line donations, and much more!) We have successfully taken something my principle…people will participate and the event will flourish.

More details on the 2013 Play for Gaucher charity poker tournament can be found by visiting www.childrensgaucher.org.

If you have questions regarding the 2013 Play for Gaucher charity poker tournament, please contact playforgaucher@gmail.com.

In this issue, we decided it would be important to outline the Three Legs of the Stool. In each newsletter we try to give an update as to the impact on research, and an eventual cure?

Our conclusion is that without the building blocks in place the “method to the madness” – meaning, is there a strategy to the efforts of the CGRF. To date, thanks to hard working people who believe in and support our mission, we have invested more than $2 million toward research – in the grand scheme of finding a cure for Gaucher disease.

Have the initial Building Blocks of Understanding the disease? If not, it is difficult for a scientist to feel confident that their thesis is valid. Since 2001 the CGRF has devoted resources to pursue a particular disease. The University of California, Davis has laid many bricks on the road to a cure for Gaucher disease.

So what are the “Three Legs of the Stool”? By doing this we are giving research scientists a “Helping Hand”. Jesus will hold a small piece of the puzzle. In this issue, we would like to recognize and thank Jesus for “Working his Magic” and lending his incredible talent to design these newsletters. I think you will agree he does a terrific job – and we are happy to offer tips and consultation on creating a website, to not give up and keep at it! Feel free to contact us at playforgaucher@gmail.com if you have questions regarding the 2013 Play for Gaucher charity poker tournament.

Simply put: if you send your hard earned dollars to the Children’s Gaucher Research Fund – It ALL goes to medical research.

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.

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All family stories can be read on the website.

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