Winter 2011–2012

HELPING HANDS

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

THE ONLY THING INCURABLE IS OUR PASSION


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Gregory Austin Macres


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History of the CGRF

In the summer of 1991 I joined the Clinical Investigation Section at the Developmental and Metabolic Neurology Branch, National Institutes of Neurological Disorders and Stroke, at the National Institutes of Health in Bethesda, Maryland. I had the honor of working with Dr. Roscoe Brady, the world's foremost expert in Gaucher disease. In our clinic we saw families from around the world who had children with neuronopathic Gaucher disease; a progressive, and more often than not a fatal disease. Families would arrive on Monday, and the children would endure five days of evaluation and testing. This was an investigative clinic, we had no cure, but the families understood that their child was receiving the best medical care possible, and that their participation was helping to further our understanding of this debilitating disease.


Greg and Deborah Macres with their daughter Ashley, and their son Gregory.

A New Family Arrives

In February of 1994, a family from California had arrived with their 1-year-old son for their first visit at the NIH. As I interacted with this family that first week I was impressed with their proactive nature. They had many questions – many of which were answered well into the evening hours after a long day of testing. Although I would arrive home late on those evenings, I did not mind because the questions were intelligent and thoughtful. To my surprise, prior to each subsequent visit I would receive from this family a Federal Express package – whereby the questions were sent in advance. Eventually they asked two questions that were very difficult to answer.

1. How is the disease affecting Gregory's brain?
2. Is anyone doing research to find a cure?

To the first question I answered "we do not know" and to the second I answered "not really … any serious research". I knew this was disheartening news, but at the time it was the truth.

Gregory Passes Away … The CGRF is Born

Greg and Deborah Macres lost their son Gregory to Gaucher disease in April of 1997. I was pleased that subsequent to Gregory's passing the family continued to stay in touch, and often discussed the possibility of establishing a charity to fund research on Gaucher disease. I was supportive, but withheld my enthusiasm understanding that this family was still coping with the grief of losing a child.

Thinking back to those phone calls some 12+ years ago I never imagined that this desire, spawned by the devastating loss of a son, would result in the Children's Gaucher Research Fund and the progress they have made over the past 10 years. The Macres family and other families from around the country began raising funds, and in 2001 they found their first opportunity to fund serious research on neuronpathic Gaucher disease. It was in the form of a newsletter I sent to the Macres's from the Weizmann Institute of Science in Israel. In that newsletter there was a small article written by Dr. Tony

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Futerman indicating that they had formulated a theory, and his laboratory had a keen interest in investigating their theory as to how the brain is affected in children with neuronopathic Gaucher disease. Prior to this I had not met Dr. Futerman, but that year I visited his laboratory in Israel. I was impressed with his facility, and to my surprise found that we had similar views as to what might be causing brain disease in these children.

> "I never imagined that this desire, spawned by the devastating loss of a son, would result in … the progress they have made over the past 10 years."

Ten Years of Work … Ten Years of Progress

Over the past ten years the CGRF has funded Dr. Tony Futerman's laboratory, resulting in very important research that has been published in nine separate scientific journal articles. Today, the answers to those two difficult questions sited above have very different answers. Because of the efforts of the CGRF we are significantly closer to understanding how the brain is affected, and yes, there is now serious research being pursued. Additionally, the CGRF recently funded the creation of an Inducible nGD Mouse Model; if successful this will significantly advance serious research in coming years.

Rare Diseases and Families

Often rare diseases do not attract interest or funding, and frequently sit idle for many years with little progress. Over the years however, families have made great contributions to medical research on rare diseases. It usually comes in the form of funding basic scientific research, thus creating the building blocks of understanding. Once these building blocks of understanding are put in place the disease then attracts more interest, and more funding for research. The CGRF has done just this – funding basic science – resulting in publications in scientific journals – sponsoring three international scientific conferences with a fourth scheduled for 2012 – the funding of the Indiclble nGD Mouse that will be available for use by laboratories around the world – links that have been made with some of the 26 other lysosomal diseases that affect the brains in children – the potential connection to Parkinson's.

Those Two Questions … Now Have Different Answers

Thanks to those of you who support the efforts of the CGRF, the answers to those two questions sited above are markedly different than they were on that day at the NIH; when a mother and father were simply trying to understand how they could best help their son. We believe, because of your support, that the building blocks are in place and that a new phase in nGD research is around the corner.

Raphael Schiffmann M.D., M.H.Sc.

Baylor Research Institute
Chairman – CGRF Scientific Advisory Board

Dr. Schiffmann grew up in Israel and served in the Israeli Army for over 3 years. He then attended the University of Liège, Belgium where he obtained his M.D. degree cum laude in 1980. He completed training in pediatrics at the Shaare Zedek Medical Center in Jerusalem, Israel and in pediatric neurology at the Boston City Hospital and Tufts New England Medical Center Hospitals. He is specialty board-certified in both pediatrics and pediatric neurology. After 2.5 years as a staff pediatric neurologist at Hadassah University Medical Center, Jerusalem, Israel, he joined the National Institutes of Health in 1991. Between 1996 and 2007 he was the Lead Investigator directing the clinical research efforts of the Developmental and Metabolic Neurology Branch at the NIH. Working at the NIH in the group of Dr. Roscoe O. Brady, who is a world expert in Gaucher disease, led to Dr. Schiffmann's interests in lysosomal diseases. In 2000, he received the degree of Master of Health Science in Clinical Research from Duke University. Since 2008 Dr. Schiffmann has been the Director of the Institute of Metabolic Disease at the Baylor Research Institute in Dallas, Texas.

CGRF Milestones

1993

- Founders son Gregory born
- Gregory diagnosed with nGD

1997

- Gregory passed away
- Memorial fund established

1997-1999

- Surprising growth of memorial fund

1999

- Children's Gaucher Research Fund established as 501(c)(3) nonprofit
- Received encouragement from National Gaucher Foundation and other Gaucher associations

1999–Present

- Attracted thousands of people to our network
- Funded seven important nGD research Projects
- Funded creation of an Inducible Gaucher Mouse
- Hosted three international medical conferences
- Raised over 1.8 million dollars for medical research

2012

- Continued funding of scientific research
- Conference of world experts: Pathological Mechanism in Neuronal forms of Gaucher Disease - Recent Discoveries – Future Direction
- Evaluation of the Inducible nGD Mouse Model

Sweet Willow

Willow Ansley Jaynes

Willow Ansley Jaynes was born on August 10, 2009 at The Med in Memphis, Tennessee. We found out something was wrong with our precious baby one week before I delivered. That was one of the most horrific moments a parent can ever imagine. The night she was born I only saw her briefly after an emergency C-section. She was polka dotted, which we would later find out is called petechia. We had a 9 day stay in the Neonatal Intensive Care unit. Multiple tests were run trying to figure out what was wrong with our sweet Willow. She was tested for CMV virus, histiopsytosis, and toxoplasmosis, just to name a few. On our 9th day there her team of doctors came to me and told me we were being transferred to St. Jude Hospital. All the tests they had run had come back negative and they thought she either had leukemia or neuroblastoma. Two horrible cancers that I prayed my daughter did not have.

> "…we found out most babies diagnosed that young were definitely going to die, usually before 2 years of age."

We were transferred to St. Jude by ambulance, and when we arrived everyone was very nice to us. She was scheduled for a bone marrow biopsy the next day. For those of you who do not know, a bone marrow biopsy it is where they drill into both hips to extract marrow from the bones. Our daughter underwent this procedure at 10 days old. The test came back inconclusive, but with concerning results. She would later undergo two more bone marrow biopsies, a liver biopsy, and a skin biopsy (from the top of her head). The first time we had to put her to sleep for a procedure was stressful, but the worst part we would come to figure out later; when your precious baby cries because she is hungry and you cannot feed her because they are NPO (no feeding by mouth). This is one of the most heart-breaking moments of a parent's life.

> "I learned how to administer her meds 3 times a day by IV … change her central line dressing, pull blood, and test her sugar."

Willow was finally diagnosed with Gaucher's Disease at 6 months of age. The first thing we did was Google it. That was heart wrenching; we found out most babies diagnosed that young were definitely going to die, usually before 2 years of age. Gaucher's affected her hemoglobin (red blood cells), platelet count, growth, development, neurological, liver, spleen, swallowing reflexes, and her respiratory. We prayed that our daughter would be one of the lucky ones to beat the disease. We kept her at home never exposing her to the world. She did not get any kind of illness until she was 7 months old. She then got bronchiolitis which put us in LeBonhuer Hospital for a month. While we were there numerous other problems were uncovered. By the time we left the hospital she had a central line in her chest and she was not able to eat anything by mouth. It was very scary coming home, but I learned how to administer her meds 3 times a day by IV, hook her up to her IV daily, change her central line dressing, pull blood, and test her sugar. She was connected to an IV pole for 17 hours a day.

Willow never got to taste things like ice cream, milk, or applesauce. She never rolled over, sat up, learned to crawl or walk. We were home for 5 months and everything was going good. She had made it to 11 pounds, into size 2 diapers, and 6 month clothes. We were scheduled for a swallow test (to see if she could go back to eating by mouth), at her one year check-up, and one year pictures. Willow was one of the happiest babies we had ever seen. Despite all the horrible things she had to undergo she always had a smile on her precious face. We woke up on August 10, 2010 ready to wish our baby girl a Happy 1st Birthday and found that she had passed in the night. Our worst fears had come true. We were shocked; she had been doing so well. Instead of planning a birthday party my husband and I had to plan a funeral. Rather than shopping for party dresses, cake, and ice cream, we shopped for a casket, funeral home, and a burial plot.

The past year has been very hard for my husband and I and our family. We are very lucky and blessed to have a great support system behind us. I think the only thing that keeps us going is knowing that she is resting comfortably in the arms of the Lord and she no longer knows sickness. We miss our sweet Willow and cannot wait to be reunited with her one day!

Jason and Amber Jaynes
Jackson, Tennessee

2ND Annual Willow Jaynes Memorial Dice Run

When our daughter passed away we knew that we wanted to do something to honor her and keep her legacy alive. We also wanted to be able to help bring community awareness about the disease our daughter suffered from. With it being such a rare disease many people have never heard of it. We decided to start an annual motorcycle ride in her memory. We have done this the past two years. In doing this event we have reached out to many local media outlets to help us to spread the word. Our event each year features a motorcycle dice run, a silent auction, a raffle, and selling bar-b-que plates. We have been very blessed with being able to put these events on with 100% of the donations from the community. Many people in our community are now familiar with Gaucher Disease and my daughter's story because of these fundraising events. We donate the proceeds to The Children's Gaucher Research Fund each year. We hope to be able to continue this event every year.

The CGRF encourages all families who have lost a child to create one annual fund raising event that honors their child's life, their struggle, and their memory.

2012 Conference

Hosted by the Children's Gaucher Research Fund

Raphael Schiffmann M.D., M.H.Sc.
Baylor Research Institute
Dallas, Texas

Tony Futerman Ph.D.
Weizmann Institute of Science
Rehovot, Israel

Pathological Mechanism in Neuronal forms of Gaucher Disease
Recent Discoveries – Future Direction

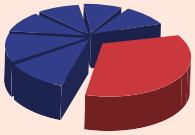
Dr. Raphael Schiffmann (Chairman of the Scientific Advisory Board) and Dr. Tony Futerman (Weizmann Institute of Science) will assist the CGRF in organizing a Scientific Conference in Atlanta, Georgia for May, 2012. For the following reasons it is believed that this is an important time to bring together the world experts.

The CGRF began funding scientific research in 2001. This, combined with the availability of the Karlsson mouse (existing mouse model developed in Europe that only lives 2-3 weeks), has provided for advances in the understanding of pathological mechanisms at play in neuronal forms of Gaucher disease. In November, 2008, the CGRF funded the creation of an Inducible nGD Mouse Model. We are hopeful that this mouse will be successful and if so, it will be available to academic and for-profit institutions around the world. We believe that the availability of this Inducible nGD Mouse Model, that lives longer than current mouse model, will pave the way toward increased research interest as well as the ability to more easily test potential therapeutic interventions. We believe that a new phase in nGD research is around the corner, and that this is the time to assemble the key players to discuss where the field currently stands, and more importantly to discuss the future direction of research. The aim of the conference is to advance our understanding of basic pathological mechanisms, but with a view to paving the way for novel therapeutic options.

Researchers in attendance will represent the fields of:

1. nGD Basic Research
2. Neuropathology of other Lysosomal Storage Diseases (LSD)
3. Non-LSD Neuropathology (i.e. Parkinson's, etc.)

Those in attendance will come from around the world, and from a wide variety of related fields. Together, from the scientific vantage point of their specialty, they will assess what progress has been made to date, and what avenues are important to pursue in the future.



2010 - CGRF Financial Audit

In the summer of 2011, the CGRF underwent its third annual financial audit. Although expensive, the financial audits allow us to participate in the Combined Federal Campaign (CFC) as well as the State employee campaigns in California, Illinois and Virginia. Each year all United States federal employees around the world as well as the employees of these three states have the option to choose the CGRF as their charity of choice in this annual fund raising drive. The campaigns take place in the months of September through December of each year.

For three years in a row the CGRF has provided these financial audits, has been accepted into all four of these fund raising campaigns, and has been awarded the "Best in America Seal".

Best In America Seal

The CGRF has recently earned exclusive recognition with the award of the "Best In America Seal". Below is the description of this honor as represented on the Children's Charities of America website:

"The Independent Charities Seal of Excellence is awarded to the members of Independent Charities of America and Local Independent Charities of America that have, upon rigorous independent review, been able to certify, document, and demonstrate on an annual basis that they meet the highest standards of public accountability, program effectiveness, and cost effectiveness. These standards include those required by the US Government for inclusion in the Combined Federal Campaign, probably the most exclusive fund drive in the world. Of the 1,000,000 charities operating in the United States today, it is estimated that fewer than 50,000, or 5 percent, meet or exceed these standards, and, of those, fewer than 2,000 have been awarded this Seal."

> "Of the 1,000,000 charities operating in the United States today … of those, fewer than 2,000 have been awarded this Seal."

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:
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