

Summer 2012

HELPING HANDS

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

THE ONLY THING INCURABLE IS OUR PASSION



We Are Often Asked...

DON'T YOU GET TIRED — Well yes, this effort with the CGRF takes a large amount of time and energy, and tired we get. The better question is how we are energized to continue this effort. To that there are several easy answers – It is our donors who believe in the cause and support the CGRF with their generous donations, and the volunteers who give of their time. There is another thing that encourages us to continue, and that is the scientific progress that is being made – this became abundantly clear at the recent Brain Conference in Atlanta.

ARE YOU CLOSE TO FINDING A CURE — My first response is that we do not care who finds the cure, so long as we (CGRF Funding) have made a direct contribution to the knowledge and discoveries that lead to that cure. I will paraphrase Dr. Raphael Schiffmann who says that rare diseases frequently sit idle for many years with little progress. By funding basic scientific research the CGRF is creating the building blocks of understanding. Once the building blocks are in place this attracts more interest, more funding, more research. The CGRF is creating the building blocks – this became abundantly clear at the recent Brain Conference in Atlanta.

TEN YEARS OF WORK...TEN YEARS OF PROGRESS — If you recall we funded the creation of an Inducible Neuronopathic Gaucher Mouse. In short, it is delayed but we believe it is going to be successful in the next 12 months. If it is, it will be freely available to any scientific research lab in the world – and they are excited and anx-

iously waiting to use it – this became abundantly clear at the recent Brain Conference in Atlanta.

WHAT ABOUT THE OTHER LYSOSOMAL DISEASES

— Yes, there are 26 other Lysosomal Diseases that affect the brains of children, and the building blocks that your donations are building are helping with understanding these diseases – this became abundantly clear at the recent Brain Conference in Atlanta.

IS THIS PARKINSON'S CONNECTION REAL

— Five years ago I would have said “I don't know but it is interesting, and I certainly would not mind if there was a connection”. At the recent conference I cornered a Parkinson's Scientist and asked him “Why are you here?” He said “I need to learn about your research to better understand Parkinson's disease”. He went on to say “If you come up with a drug therapy for children with neuronopathic Gaucher disease, we will, unquestionably, give it to Parkinson's patients”. As you can imagine, I was a bit floored. I believe it is safe to say, yes, there is a connection between Gaucher disease and Parkinson's – this became abundantly clear at the recent Brain Conference in Atlanta.



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

Inside
2012 Brain Conference

A Meeting of Minds

Valerie Yannias
The Karate Queen
is sixteen!

-continued from last page

WILL PARKINSON'S RESEARCHERS USE THE CGRF'S MOUSE

— YES - this became abundantly clear at the recent Brain Conference in Atlanta.

- 100% of your donations go to fund Medical Research.
- We believe that the research you are funding is making a huge impact
- This became abundantly clear at the recent Brain Conference in Atlanta.

The Only Thing Incurable is Our Passion

WHY WOULD A PARKINSON RESEARCHER COME TO A GAUCHER'S MEETING?

By John Hardy PhD, FMedSci FRS

I work on Parkinson's disease which affects half a million Americans... including, Michael J Fox, Mohamed Ali, Janet Reno as well as Pope John Paul II. Gaucher's disease, in contrast, affects between 5,000 and 10,000 Americans.

The answer is simple: 5 years ago, scientists, led by Ellen Sidransky at the National Institutes of Health, found that some cases of Parkinson's disease were predisposed to by the same defective gene, glucocerebrosidase, that causes Gaucher's. This tells us all that something about the causation of the two diseases is similar. So the answer is... I came to learn!! Gaucher's researchers have been studying this gene for years and have learnt a lot about its function in cells, and how loss of that function damages cells and can kill them. I want to understand what they already know.

I also came to teach Gaucher's researchers what I know about Parkinson's. We Parkinson's researchers can learn a lot from the enormous amount of knowledge Gaucher's researchers have understood about their (your) disease and we can also explain what we already know about Parkinson's disease and see how much of that knowledge might be useful for Gaucher's disease. Perhaps most

useful, we can also help bring much needed money into Gaucher's research. Before scientists realised the two diseases were related the amount of research effort into Gaucher's was very small because it affected so few people...now, the

"I work on Parkinson's disease which affects half a million Americans...including, Michael J Fox, Mohamed Ali, Janet Reno as well as Pope John Paul II."

amount of research into Gaucher's is becoming more well-funded...and, this is desperately needed. There is so much we still do not know about both diseases. In particular, we really don't understand why in some people Gaucher's disease is so mild and in others so severe even if they have the same genetic defect. For us to understand this we need to study the disease in hundreds of Gaucher's patients and this will cost a lot of money...perhaps the fact that this knowledge will also benefit the hundreds of thousands of people with Parkinson's disease will help us get the funding we need to do this.



JOHN HARDY PHD, FMedSci FRS

Reta Lila Weston Research Laboratories,
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London, England

Brain Conference

ATLANTA GEORGIA – May 18, 2012

TONY FUTERMAN PH.D.

Weizmann Institute of Science
Rehovot, Israel



As many of you know I have been working on neuronopathic Gaucher Disease for over a decade in my lab at the Weizmann Institute in Israel. We have made great progress creating the "building blocks" of understanding. This progress has happened because of the support received by the CGRF – or more personally – because of your generous support. However, one of the frustrating aspects with research in this area is the small number of people who have been working on this disease. The meeting that was held in Atlanta brought together in an informal setting 25 brain experts from around the world, some of whom had previously worked on Gaucher Disease and a number who were totally new to the field but work on various aspects of brain diseases. Getting this group together was of enormous importance, as it is sure to foster new collaborations and new ways of thinking about the disease.

The meeting was very informal, inasmuch as the scientific sessions were centered around discussion points which had been selected ahead of time. Participants were encouraged to suggest the most outlandish ideas, to participate freely and to engage in lively debate. These goals were achieved and I think it would be fair to say that this was one of the most intense two day discussions that I have ever participated in! It was particularly encouraging to get the input of scientists who did not come from a Gaucher background and many of them had suggestions which, I believe, will be of enormous benefit to the field.

I can do no better than quote one of the participants who

"It was particularly encouraging to get the input of scientists who did not come from a Gaucher background and many of them had suggestions which, I believe, will be of enormous benefit to the field."

sent me an e-mail after the meeting and said something along the lines of "I was skeptical that a meeting like this could work, but in the end I think it was fantastic". I hope that there will be another similar gathering in the near future to brainstorm about progress made during this period and to inspire new ideas and new research directions.



A Meeting of Minds ATLANTA 2012

Unfortunately, it is a familiar story to those of us who study rare genetic disorders – lack of funding. Neuronopathic Gaucher disease is also a victim of this dire situation. For those few who are fortunate enough to have the resources to study this disease, it is often a lonely and secluded existence. We are scattered in hospitals and laboratories around the world doing the best that we can with finite resources and limited budgets. When faced with these hurdles, communication between us is critical in terms of sharing ideas, data, expertise, establishing collaborations and avoiding wasteful repetition. Although the world is becoming smaller in terms of our ability to communicate, nothing quite equals the experience of a good face-to-face discussion.

It was for exactly this reason that I was so pleased to be invited to attend a meeting organized by the Children's Gaucher Research Fund in Atlanta, May 2012. Under the Chairmanship of Professors Tony Futerman (Weizmann Institute, Israel) and Raphael Schiffmann (Baylor Institute, USA), experts in the fields of Neuronopathic Gaucher disease and other closely related neurological diseases were asked to attend. Furthermore, representatives from industry were also present. The format was uncon-

“The clinicians amongst us have the vital role of translating any therapeutic advancement from the bench to the clinic...all the players required in this sequence of events were present in Atlanta and this does not happen often.”



ventional in that this was not typical of a conference. Rather, it had the feel of an informal lab meeting with all present sitting around a single table. It reminded me of the old Arthurian Legends in which all Knights of the realm sat as equals with the King within Camelot. In a similar fashion, everyone from Professors down to PhD students were free to speak, criticize, encourage and present their views and data. The discussions were open, frank and to the point. I found it both refreshing and enlightening.

“What was clear to me is that significant advancements are being made in our understanding of (Gaucher) this terrible disease.”

A broad variety of subjects were raised by attendees including: gene therapy (Dr Ahad Rahim, University College London, UK), other alternative therapies (Dr Seng Cheng, Genzyme, USA), disease pathophysiology in mouse models (Dr Einat Vitner and Ms Tamar Farfel-Becker, Weizmann Institute, Israel), biochemistry and cell biology (Dr Greg Grabowski), clinical perspectives (Dr Raphael Schiffmann, Baylor Institute, USA and Dr Ashok Vellodi, Great Ormond Street Hospital, UK), the link between genotype and phenotype (Dr Andres Klein, Weizmann Institute, Israel).

Three distinct presentations were also given on the association between Gaucher disease and Parkinson's disease (Dr Ellen Sidransky, NIH, USA, Dr John Hardy, University College London, UK and Dr Dimitri Krainc, Harvard Medical School, USA). The link between these diseases is attracting significant attention from research groups around the world. Research into Parkinson's disease is significantly better funded than Gaucher disease. It benefits from having a higher profile due to its more significant impact on global health in an ageing population. By association alone, this is likely to raise awareness of Gaucher disease.

Another useful feature of the meeting was the presence of experts in diseases similar to neuronopathic Gaucher disease. These included: Professor Alessandra d'Azzo (St. Jude Children's Hospital, USA), Professor Volkmar Gieselmann (University of Bonn, Germany), Professor Marc Patterson (Mayo Clinic, USA), Dr Konstantin Dobrenis (Albert Einstein

College of Medicine, USA), Professor Daniel Ory (Washington University School of Medicine, USA), Dr Morris Benveniste (Morehouse School of Medicine, USA) and Dr Emyr Lloyd-Evans (Cardiff University, UK). They provided invaluable input and highlighted both similarities and differences between the neurological diseases they

work on and Gaucher disease. It is often very easy to become very 'Gaucher-centric' and insular so objective points-of-view are welcome and we can draw from each other's pools of knowledge.

This was an excellent meeting and I came back to the UK more informed, with new ideas and contacts that will hopefully form fruitful collaborations. What was clear to me is that significant advancements

“Three distinct presentations were also given on the association between Gaucher disease and Parkinson's disease...”

are being made in our understanding of this terrible disease. This, in turn, will help scientists, like myself, design and target novel therapies that will have a tangible impact on the lives of patients. The presence of representatives from industry is vital in making such a therapy scalable and realistic from a production and economic point of view. The clinicians amongst us have the vital role of translating any therapeutic advancement from the bench to the clinic. Therefore, all the players required in this sequence of events were present in Atlanta and this does not happen often.

I sincerely hope that the CGRF will organize future meetings because this one was very useful.



DR AHAD A. RAHIM
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You fund her research

The research that Tamar Farfel-Becker pursues in the Futerman Lab at the Weizmann Institute of Science is funded by you – those of you who support the CGRF. Below are Tamar's thoughts regarding the recent conference in Atlanta, Georgia.



“Hi GREG,

Just wanted to share my feelings about the meeting - The experience of discussing science with the world experts of our field, just for sharing ideas and thinking how to promote the field was very unique, exciting and helpful. I have no doubt that new collaborations will come of it, and new players will join the field. I think that these are exactly the “building blocks” that you mentioned.”

TAMAR FARFEL-BECKER

Department of Biological Chemistry
Weizmann Institute of Science
Rehovot, Israel



Greg Macres (left) Deborah Macres (middle) Tanya Collin-Histed (right).

DEAR GREG AND DEBORAH

“Finally I am sat at my desk after a long and busy week. I wanted to send you both a message to say a big *THANK YOU* to you both for last weekend, and for inviting me to attend the meeting.

The meeting was a breath of fresh air and to have so many people in one room all talking about nGD was truly wonderful. The work you have done with Tony, and the scientists and clinicians in the US with research is truly humbling.

It was of course lovely to see you both and spend time with you. Greg I loved hearing about the journey you and Deborah have been on.

I think the outcome was a very positive and I look forward to seeing how this will unfold and go forward in the future.

Hey, keep up the good work, if there is anything I can do please ask.

With great affection

TANYA COLLIN HISTED
Chief Executive
UK Gauchers Association

Dursley, Gloucestershire, UK

The Karate Queen is 16!

By Cynthia A. Yannias

Yes, Valerie Yannias, a young person with Type III Gaucher Disease is sixteen and a Karate student. She has already won several medals and will be attending her first National Karate Tournament in July. Valerie has also climbed all 2,000 + steps of the Willis Tower (formerly the Sears Tower). Despite her physical challenges, she has pursued her dream to be an athlete. Karate has helped her with her balance and posture. We thank Sensei Jeff Kohn, Founder of Karate Can-Do for giving her the opportunity to express herself through Karate.

“It was an instant connection, a bond forged not of the womb, but of the heart.”

Valerie is remarkable! From an infant she was curious about the world around her. She stared intensely at the stuffed animal puppet show we performed daily, listening to every word, grabbing my finger at four days old. It was an instant connection, a bond forged not of the womb, but of the heart.

Sixteen years ago, we embarked on a journey that took us 8,000 miles from home to receive a precious gift from God, a daughter to love and cherish. Some people thought we were crazy to adopt a child from Greece, but family and friends were supportive, giving us love and encouragement throughout the ordeal. It may not have been labor, but it was five and half months of dealing with both the Greek and American bureaucracies! When we looked back in time, it is sometimes hard to believe what transpired. No one does anything alone. We know that our family and friends' prayers were helping us every step of the way, even to this day. Life is full of challenges and it is faith that sees one through the darkest of times. Believing in that higher power that is all knowing, loving and merciful

is the difference between hope and despair. Our challenge came when Valerie was two years old and was diagnosed with Type III Gaucher Disease, a rare, genetic enzyme deficiency that is life threatening without treatment. This form of the disease had neurological implications.

At first it was believed that she had the rarest of forms that would surely cause death by two-three years old. But a geneticist that calls himself a “country

doctor,” Dr. John Barranger from the University of Pittsburgh Medical Center, saw

in her determination, intelligence and a beautiful smile. We will not forget his words, “she will declare herself.”

Declare herself indeed! Valerie has met the challenge of living with Gaucher Disease with grace and dignity, enduring

“Sixteen years ago, we embarked on a journey that took us 8,000 miles from home to receive a precious gift from God, a daughter to love and cherish.”

spinal fusion surgery, infusion therapy every two weeks for 14 years and an overprotective mother! She has a quirky, but funny sense of humor and a sharp wit. All you have to do is meet “Sister Hand and Jane,” the talking hands who represent the voices of the good and bad conscious. They are the alter egos, whose back and forth repertoire is hilarious. Despite the disease, Valerie is your average teenage girl, who crushes on Justin Bieber, knows all the words to the latest Shake-It-Up songs and loves to shop. Valerie may be adopted, but she did inherit two traits from her grandmothers. From Yiayia Bessie she



learned the art of tchotchke shopping, from Yiayia Eleni the art of procrastination.

Karate has been a Godsend for Valerie. It has given her the opportunity to be an athlete. It has strengthened her mind and body. You don't ever want to be elbowed by Valerie! She has moved into the arena of competition with a determination to be the best she can be. Karate is such an integral part of her life. It's a struggle to get her up for school every day, but on Saturdays she is up at the crack of dawn to go to Karate!

Now our challenge is to get her through high school and onto a meaningful, productive position that will allow her to live independently. Every life has a purpose and our task is to help Valerie find hers. We're doing our best to teach her to be a respectful, caring, productive and responsible person.

We are blessed and full of gratitude to God, our family and each and every one who has touched our lives and Valerie's. Life is to be cherished, shared and celebrated. It is a gift to be nurtured and treasured. Thank God for the Karate Queen!



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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