

Summer 2015

HELPING HANDS

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



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Gregory
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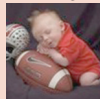
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HERE IS A SIGN OF PROGRESS

There was a time that we published a 4-Page newsletter and we struggled to fill the space. Today we publish a 6-Page newsletter and there is not enough room to share all of the exciting things that are happening. Yesterday we would publish hope... today we publish progress... tomorrow... we are quietly optimistic.

Just everyday people

This has always been a grass-root effort - everyday people who support the cause. Everyday people who have chosen the CGRF as their "charity of choice"... Perhaps because they lost a child or a grandchild... Perhaps because they know a family who has endured tremendous grief... Perhaps because they have simply never found a charity whereby 100% of their gift will go to the cause. Everyday people have collectively helped raise over \$2.5 million. We are humbled.

Scientific Breakthrough

One year ago we published details of a scientific breakthrough at the Weizmann Institute of science - research that you funded. The Board of Directors for the CGRF is encouraged by the scientific progress and has awarded the Futerman Laboratory at the Weizmann Institute of Science with a \$400,000 research grant.

A look behind the curtain (See inside) - Deborah Macres responds to Dr. Vitner

Einat ... Thank you for your beautiful journey you put into words. I have to say this is one of my all-time favorite articles that will be put in the newsletter. It touches not only on the research journey that has a time-line that involves so much

patience and determination, as well as adding your human and heartwarming journey of a woman scientist, mother, mentor, and team member. So well done you have brought tears to my eyes and warmed my heart - a connection that makes our world so much more intimate even while living on different continents. I can only imagine how proud of you your family is as you have been instrumental in this scientific journey of discoveries that help unlock the mysteries and the future of treatments. ■



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AND GREG MACRES

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RUNNING A RESEARCH LAB



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



“Einat’s passion for basic research shines through the article, as does her enthusiasm and sense of humor.”

Although science is generally considered to be an objective business, that’s to say, a bunch of people examining the facts and reaching conclusions, science is actually far more than that. Science is all about people and about generating an atmosphere where people feel able to comment and positively criticize other people’s thoughts, ideas and hypotheses. I have been fortunate

over the past decade or so to have had a fantastic bunch of people in my lab and many of them are working on neuronal forms of Gaucher disease, funded by the Children’s Gaucher Research Fund. Among these people are Dr. Tamar Farfel-Becker, who is now working as a postdoctoral fellow in the National Institutes of Health in Washington D.C., Dr. Andres Klein, who is about to take up a postdoctoral position in Naples, Italy, and last but not least, Dr. Einat Vitner, who obtained her PhD in my lab 2 or 3 years ago and has been working as a Senior Research Fellow ever since. The bad news is that Einat is likely to leave the lab in the coming months and this will leave a big hole in my research group.

Einat has written a personal and very moving account about the time that she has spent in my lab and you can read her account in the accompanying article. I think the account beautifully sums up the perspective of an excellent research colleague. Einat’s passion for basic research shines through the article, as does her enthusiasm and sense of humor. I should

“I well remember a day a couple of years ago when Einat came into my office and excitedly told me that the RipK pathway seemed to be a key player in Gaucher disease.”

like to emphasize that much of the progress that we have made over the past few years is largely due to Einat’s input and hard work — it’s often the professor who gets the glory, but in this case Einat deserves a huge amount of credit for her outstanding work. As many of you may know, we recently published what I consider to be a breakthrough discovery in the neuronal Gaucher field inasmuch as we showed that RipK is an essential player in the pathways of neuronal cell death and neuroinflammation. I well remember a day a couple of years ago when Einat came into my office and excitedly told me that the RipK pathway seemed to be a key player in Gaucher disease. At the time, I had never heard of RipK and this perfectly illustrates Einat’s contribution to the lab. I hope you enjoy reading her article. ■

A LOOK BEHIND THE CURTAIN



Einat Vitner Ph.D.
Senior Research Fellow
Department of
Biological Chemistry
Weizmann Institute of Science
Rehovot, Israel

In 2006, 9 years ago, I received my Masters in biology and was looking for a place to continue my training. The Weizmann Institute is considered the number 1 research institute in Israel, and I was naturally very excited when I was accepted.

What disease to study

When looking for a field to specialize in, one thing was absolutely clear to me: I wanted to study a disease. I was 25, naive and filled with motivation and wanted to do something that would leave a mark. In my search I found Prof. Tony Futerman. Tony had published a number of seminal papers on Gaucher disease, and his research was very appealing to me.

Actually, I had absolutely no idea what Gaucher disease was! In fact I had never heard of it before, but the combination of a chance to study a disease with limited treatment options and the

strong impression left by Tony’s research and Tony himself was enough to get me hooked and I gladly joined the lab.

When I did, the Gaucher program in Tony’s lab was not that strong, and Tony was glad of the opportunity to reestablish the focus on Gaucher research. Almost immediately after I joined the lab, the CGRF organized a scientific conference on Gaucher disease. Tony proposed that I join him so that I could learn about recent progress. I was very glad to have this opportunity to attend my very first conference, and immediately embarked on what would be my first research work in the lab: what does one wear to a conference? Needless to say I ended up stuffing most of my wardrobe into my overweight suitcase, only to meet Tony’s sarcastic smile as he boarded the plane with just a small carry-on... The conference itself was fascinating. Meeting leading researchers in the field was quite an experience. My strongest memory of the conference was the debate that revolved around the connection between Gaucher disease and Parkinson’s. There were very heated discussions, and most of the scientific community was very skeptical about this relationship.

CGRF funds our research

One of the most important things I learned in my early days in the lab was that doing science costs money. Tony got me a Macintosh and said:

“You should start clarifying to yourself exactly what you want to study because we need a grant to cover your work”. I nodded vigorously in agreement, all the while trying to figure out how to get the Mac to work, so that I could Google what a grant is and what you need it for. I then spent a few weeks reading a lot. I learned what Gaucher disease is, what was known about the disease, and, most importantly, what was not known. Tony and I brainstormed on potential research directions and put together the grant application, which we then sent to the CGRF. When the grant application was accepted, I was thrilled – we could set to work now.

The first year of my research was hard and frustrating. At that time, there wasn’t a good mouse model for the disease, and most of the work was done in-vitro (working on cell cultures in petri dishes). The work on cultures was exasperating and, sadly, not very fruitful. Luckily, at the same time Tamar Farfel-Becker joined the lab as a grad student (and later PhD student), and we spent many hours side by side on the microscope working and chatting. In 2007 the breakthrough came, when Stefan Karlsson’s lab created a mouse model of the disease and was kind enough to share this mouse with us. This model emulates the neuronal impact of the disease in live mice. This advance opened up many opportunities for research that had been impossible until that point.

“REHOVOT SCIENTISTS BRING HOPE OF AN EVENTUAL TREATMENT FOR CRUEL, INCURABLE FORM OF GAUCHER'S DISEASE...”

Jerusalem Post

CGRF Conference

In 2008, the CGRF organized another conference on Gaucher disease. By then we had become a “Gaucher group” in the lab, and we all went to the conference. It was then that I had the pleasure to personally meet Greg and Deborah Macres. This conference was a major crossroads in my PhD, and had a tremendous impact on me for several reasons. The lab is a kind of “bubble”. In the positive sense, this bubble protects you from the outside world and allows you to concentrate on your questions, research and work. Although we study a disease, I had never actually met a patient until that point. When I met this couple, things started connecting, and everything became very real. Sentences I wrote in my thesis such as “If we understand the pathway leading to neuronal loss in Gaucher disease we may be able to find new therapeutic directions” came to life. At dinner, Greg shared his moving story, and that of the foundation, and there was not a dry eye left in the audience.

As opposed to the previous conference, in this one, scientists began demonstrating an undeniable connection between Gaucher and Parkinson's. What was heatedly disputed two years earlier was becoming acknowledged as a fact. It was at this conference that I also presented the preliminary results of my research for the first time. This was the first time I ever gave a scientific lecture, and I was naturally very excited. After the presentation, Deborah approached me, hugged me and said: “I have no idea what you were talking about, but that was great...”. She thanked me for studying Gaucher. Needless to say, I was very moved, and went back motivated and filled with new ideas. It was also around this time that my research started to pick up.

Step by step, we analyzed the mouse model of the disease, and learned a lot about the different stages of disease progression. We realized that inflammation and the immune system have a fundamental role in disease progression and neuronal damage. We understood which regions of the brain are damaged, and started understanding which proteins are involved in disease progression.

Break Through

One of the proteins whose involvement we noticed is called ripk3. The most exciting experiment we conducted was testing whether the inhibition of ripk3 can slow down disease progression in mice. Every day when we came over to the mouse cages to watch their behavior, our hearts raced. When we saw that mice without ripk3 lived longer and got better we almost couldn't believe it. Perhaps our research would end up with a significant scientific contribution after all!

“WEIZMANN INSTITUTE FINDINGS MAY POINT THE WAY TO A TREATMENT FOR A DEVASTATING CHILDHOOD DISEASE...”

Health Canal

In 2012, the GCRF arranged another conference, in Atlanta this time. It was a special and different conference than those that had been done in this field so far. It was a round table conference: a group of scientists, from all over the world, brainstorming on Gaucher disease. Tony and I shared our exciting findings with Greg. I remember Greg asking questions, trying to understand every detail.

Break Through Published

When our paper on the subject was accepted by the prestigious journal, Nature Medicine, we were all overjoyed. Beyond the contribution to understanding the disease and the hope to find a treatment, the paper has had a significant

contribution in exposing the broader scientific community to Gaucher disease. This in turn can drive more research and will hopefully accelerate the path to understanding and treating this disease. I think one of the most significant developments in the years I have been studying this disease is the fact that it is more broadly recognized today, and the number of labs working on it has grown. The relationship between Gaucher and Parkinson had a major contribution to this and is fueling more research of Gaucher's disease.

My mom's a Scientist

My eldest son, Gilad, is now 5 years old, and loves coming to the lab to help me whenever he can. Last week, when I came to the kindergarten to pick him up, his teacher told me my son had a “very developed imagination”. When I asked why, she said: “He said his mother is a scientist and studies Gaucher disease” (I guess the fact that I wear jeans and a t-shirt when I drop him off and chase around two hyperactive boys wasn't her idea of a research scientist!). “I wonder how he made up this ‘Gaucher disease’”, she concluded. So, I guess there is still work to be done in raising awareness of this disease among kindergarten teachers, but on the scientific community front, things are much better...

A Personal Note

Recently, I have been thinking a lot about Greg and Deborah Macres. Two years ago my father, to whom I was very close, was diagnosed with advanced prostate cancer. The disease was discovered at a late stage and, unfortunately, the currently available treatments were not able to stop its progression. At first I could not avoid the thought that had I only chosen to study cancer, maybe I could have helped more, understood more, known the right people... Of course, it is naïve of me to think so, but as I stood there helpless, I often thought of Greg and Deborah, and my appreciation for them grew even more: the way in which they chose to cope and the special bond between parents and children. I hope I can be a bit more like them. ■

“GAUCHER PATIENTS OFFERED NEW HOPE...”

Medical News Daily

GAUCHER SPECIALISTS MEET IN BERLIN



Raphael Schiffmann
M.D., M.H.Sc.
Baylor Research Institute
Chairman – CGRF Scientific
Advisory Board



I would like to briefly report on an outstanding meeting which I attended in Berlin at the end of March 2015. The meeting was sponsored by Genzyme and was the sixth in a series of meetings known as the Gaucher Leadership Forum. On this occasion, Professor Tim Cox from Cambridge University in England, put together an absolutely outstanding program which brought together most of the leaders in the field. Lectures ranged from subjects as esoteric as the physiology of the lysosome to the use

of new cell types to study Gaucher disease research, overviews of emerging therapeutic options, and the association between Gaucher disease and lymphoma and myeloma. But perhaps one of the most moving and impressive lectures was given by a Dr. Cheema from Lahore in Pakistan who spoke about the burden of neuronopathic Gaucher disease patients in Pakistan. Apparently due to the limited funds available to the Pakistani health system, very few if any Gaucher disease patients are able to be treated and moreover, most of the Gaucher patients that Dr. Cheema sees in Lahore have a severe Type 2 form of disease; thus even if a treatment was available for Type 1 Gaucher disease, this would not be of any help to these severely affected children. Dr. Cheema's talk brought home the human aspect of Gaucher disease and for those of us who are basic scientists, made us realize that the need for finding treatment options for neuronopathic Gaucher patients is pressing. ■

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FUND RAISING IS THE KEY TO PROGRESS

Progress made by the Children's Gaucher Research Fund would not be possible without our generous donors, and without the commitment of families who have lost a child to neuronopathic Gaucher disease. Many families choose to honor their child's life by fundraising and supporting our research effort.

Play for Gaucher – Charity Poker Benefit

"Play for Gaucher" (pronounced Go-Shay) was established as an annual event to raise money for the Children's Gaucher Research Fund...a non-profit [501(c)(3)] organization that provides grants for medical research to find a cure for the fatal infant variant of this disease, along with other childhood genetic and brain disorders. Currently, there is no cure

for Gaucher's Disease and children diagnosed typically pass away before the age of 2



On July 13, 2009, Gaucher's Disease claimed the life of a very special little boy...Lincoln Lewis Wheatley. We hope to not only raise awareness of the disease, but also raise money for the medical research fund and pediatric research in general. Hopefully our efforts will allow

researchers to learn more about this and similar genetic diseases - and bring us one step closer to finding a cure! Although forever in our hearts, this event is held in Lincoln's memory. Lincoln was 8 months old at the time of his passing, but his spirit remains with us forever. ■

Zachary and Kelly Wheatley
Powell, Ohio



KEY-CLUB'S ROCK!

Pleasant Grove High School KEY CLUB brings together Sacramento area High Schools for their 2015 Awake-A-Thon benefiting the Children's Gaucher Research Fund. It is amazing. Over the last 10+ years High School students have joined hands... lending a "helping hand"... raising over \$75,000 for Gaucher research.



HEAVEN BIRTHDAY RIDE

Each year the Macres Family celebrates little Gregory's Heaven Birthday with a charity Bike Ride visiting important landmarks in his life. The only riders are Gregory's Mother, Father, Sisters, and Brother – who are all sponsored by friends & family resulting in an event that honors Gregory's life, and again raises close to \$5,000.





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