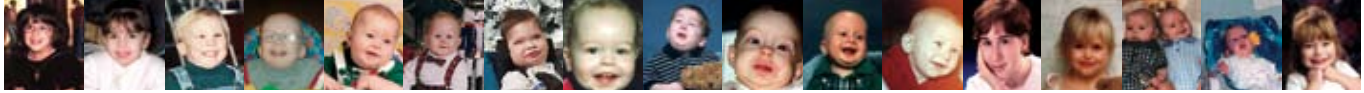


# HELPING HANDS

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



## THE ONLY THING INCURABLE IS OUR PASSION.



Valerie Yannias	Victoria Casares	Gregory Austin Macres	Noah Jerome Shaffer	Kyle Herrel	Jared Patrick Ashley	Joseph DeFacci	Andrew Dennis Doran Jr.	James Tyler Cooper	Lauren Marsh	Ryan James Conklin	Cameron Robert Watson	Kristina C.	Madeline Collin	Grant & Garrett Geyer	Hannah Colwell	Emma C. Pozzobon
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*Greg Macres*  
**Gregory Macres**  
Chairman/Founder



*Deborah Macres*  
**Deborah Macres R.N.**  
Founder

Each of you who support the CGRF are making an enormous contribution to medical science. There is no doubt that one day the consequence of your compassion will be felt by parents and children around the world. The progress that has been made became abundantly clear at the Lysosomal Diseases and the Brain conference that was held in June 2006. Over 100

medical scientists from ten different countries assembled to discuss research that has never been discussed before. A few short years ago assembling this group of world-renown scientists, much less the science that was discussed, would have seemed impossible. We may still be in the beginning stages of finding a cure, but the momentum is clearly building. You are winning "the

*“To laugh often and much; to win the respect of intelligent people and the affection of children; ... to leave the world a little better; whether by a healthy child, a garden patch or a redeemed social condition; to know even one life has breathed easier because you have lived. This is the meaning of success.”*  
– Ralph Waldo Emerson

respect of intelligent people", and you are winning "the affection of children". Read Tanya's words on page 2 of this newsletter; a mother who is intimately involved, and a daughter who is "Wishing Upon a Star". Your collective efforts will undoubtedly leave the world a better place. "This is the meaning of success."

## The CGRF Funds 3 Years of Research

In 2003 and 2004, with your hard-earned donations, the CGRF funded two years of research with Dr. Tony Futerman at the Weizmann Institute of Science in Israel. The research focused on determining the mechanistic relationship between defective Ca<sup>2+</sup>-homeostasis via the ryanodine receptor (RyaR) and the type of Gaucher disease, i.e. types 1, 2 and 3.

Layman's Translation:

1. On a molecular level, how does the disease affect the neurons in the brain?
2. Why is the brain affected in type 2 & 3 Gaucher disease, but not affected in type 1 Gaucher disease?
3. Does the regulation of calcium, which naturally occurs within neurons, become defective, thus impairing the function of neurons in the brain?

The results of this research were published in the scientific literature in early 2005. Subsequent to this publication seven other relevant papers that relate to LSDs and/or defective calcium have been pub-

lished by a collaboration of Dr. Futerman's laboratory and others. Of great interest is the fact that defective calcium-homeostasis has now been reported in a number of lysosomal storage diseases, including Gaucher type 2, Niemann-Pick A, Sandhoff disease, and the GM1 gangliosidosis, and in a number of other lysosomal storage diseases

*“... the mechanisms first described in type 2 Gaucher disease (research funded by your donations) may be of relevance for understanding other LSDs, and possibly other neurological diseases.”*

(LSDs), implying that the mechanisms first described in type 2 Gaucher disease may be of relevance for understanding other LSDs, and possibly other neurological diseases.

The CGRF has recently funded a 3-Year Research Grant with the Weizmann Institute of Science to study up-and-down-stream pathways that may be altered in neuronopathic forms of Gaucher disease.









