

Award Granted to Prof. Mia Horowitz of Tel Aviv University in Memory of Ellie Carter

In the December 2006 edition of the Gauchers News the Association invited research applications in the area of Type 2 Gaucher disease for consideration.

The Association are pleased to announce that after three applications were considered a three year grant has been awarded to Prof. Mia Horowitz of the Department for Cell Research and Immunology, Tel Aviv University, Israel. The project will contribute towards the understanding of the molecular mechanism underlying brain cell death in neuronopathic Gaucher disease.

The funds to support this research have been raised by the family of Ellie Carter who died on 9 February 2004 aged seven months. Ellie had Type 2 Gaucher disease. In memory of Ellie her parents Jill and Ian, set up ELF, the Eleanor Lily Foundation, to raise funds

to go towards research into understanding more about Type 2 Gaucher disease.

In accepting the research grant Prof. Mia Horowitz said "I was delighted to realise that I was the recipient of this award from the UK Gaucher association, which will support our efforts toward understanding the molecular mechanism underlying brain cell death in neuronopathic Gaucher disease. As part of this project, we intend to study what are the consequences of the endoplasmic reticulum associated degradation of the mutant glucocerebrosidase forms. We believe that the accumulation of mutant glucocerebrosidase forms in the endoplasmic reticulum of Gaucher patients with neuronopathic forms of the disease lead to unfolding protein response and cell death. We would like to test this hypothesis. During these



Ellie Carter

processes mutant glucocerebrosidase is supposed to interact with several proteins, which we would like to unravel."

Research at the Weizmann Institute in Memory of Emily Downes

Emily Downes died in 2003 aged one year. Emily had Type 2 Gaucher disease. Funds in Emily's memory to be used towards research into understanding more about Type 2 Gaucher disease.

The Gauchers Association has now awarded Prof Tony Futerman of the Weizmann Institute, Israel a grant in memory of Emily to further his research program into understanding the pathological mechanisms at work in the brains of type 2 patients, and in particular, in nerve cells.

Prof Tony Futerman said; 'There has been some dispute in

the field about whether nerve cells are themselves affected in type 2 Gaucher, but we have provided data over the past few years showing defective calcium homeostasis in neurons from animal models of type 2 GD and also in human brain (obtained post-mortem). We are continuing to define the biochemical pathways that are



Jeremy Manuel pictured on a recent trip to Israel with Prof. Tony Futerman and Einat Vitner a PhD student working in Prof Futerman's laboratory funded by the Children's Gaucher Research Fund

affected as a result of these changes in calcium homeostasis, and this award will help us to pursue these studies, with a view to eventually identifying novel therapeutic approaches'.