The Publication of Gaucher Disease edited by Prof Ari Zimran and Prof Tony Futerman

Gaucher disease has proven to be a paradigm for many lysosomal storage disorders. The scientific research and clinical management of the disease has been ground breaking not only for Gaucher patients but also for patients with other such disorders.

Gaucher disease was the first disorder to have treatment by enzyme replacement therapy and it was on Gaucher patients that substrate reduction therapy was first trialled. Gaucher disease was also the first storage disorder to be designated with national status by the Department of Health ensuring that all patients were treated at one of the four national treatment centres.

The publication of *Gaucher Disease* is another first. Edited by **Prof Anthony Futerman** from the Weizmann Institute of Science and **Prof Ari Zimran** of the Shaare Zedek Medical Centre in Jerusalem, this book is the first truly comprehensive and multifaceted reference on Gaucher disease. It details not only the most current treatment modalities and the directions of new treatment but also looks at all aspects of diagnosis, patient management and ethical considerations. Contributors include world renowned experts, patients' organisations, pharmaceutical companies, public health leaders and physicians. **Jeremy Manuel OBE, Susan Lewis** and **Tanya Collin-Histed** of the Gauchers Association are contributing authors to the chapter 'Patients Perspective'.

Such is the importance of the book that together with the UK Friends of Weizmann Institute and Shaare Zedek, the UK Gauchers Association celebrated its publication with a reception held on 20 November. Generously sponsored by a law firm S.J Berwin LLP where around 80 people had the opportunity to meet the editors.

Members of the Association can obtain a copy of the book at a reduced price. Please contact Tanya Collin-Histed: ga@gaucher.org.uk or telephone: 00 44 (0) 1453 549231.

NSCAG Designation for LSD to be extended

In its May 2005 edition, Gauchers News reported on the Dept of Health announcement that for two years, from April 2005 to March 2007, Cerezyme and Zavesca prescribed for patients with Gauchers disease was to be centrally funded by the Dept of Health under the auspices of the National Specialised Commissioning Advisory Group (NSCAG). In September NSCAG agreed to continue to commission services for the treatment of patients with Lysosomal Storage Disorders until March 2008.

The Chariman of the Gauchers Association attends regular meetings of the Expert Advisory group for NSCAG's lysosomal storage disorder service.

Update from Addenbrooke's Hospital

The Lysosomal Disorders Team is pleased to announce two new appointments to the group, writes Dr Patrick Deegan, Consultant at Addenbrooke's Hospital, Cambridge:

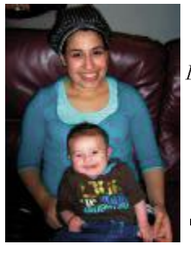
'**Sister Jane Tindall**, familiar to many of the readers of the newsletter for her involvement with the Bone Research Project, has taken up a position within the Addenbrooke's NHS Trust as a Clinical Nurse Specialist. We are very fortunate to have Jane as a valued member of the clinical team.

'**Dr Penny Stein** has been appointed to a senior research position within the University of Cambridge but very much allied to the Lysosomal Disorders Unit as a practising clinician. Doctor Stein will bring to the group a wealth of scientific research experience in addition to her excellent clinical and personal skills.

'We are also pleased to announce that **Sister Liz Morris** has taken on an expanded role in the service, with increased managerial responsibility in the running of the unit in addition to her clinical nursing role.

'Six months have passed since the opening of the new Lysosomal Disorders Unit in Addenbrooke's Hospital. We hope that our patients are as pleased with the facility as we are. We are now able to provide nursing, medical and secretarial services in a single, compact suite of rooms. Most of the essential materials for running a clinic are close to hand, like patients' notes, fax machine and tea.

'A particular advantage is the ability to give infusions during the outpatient clinic. The facility gives us more flexibility to see patients outside of normal clinic times for particular specialist investigations and treatments. We look forward to seeing you there.'



Congratulations to Shevi and David on the birth of their second child Zacharia who was born on 17 July 2006. Shevi has Gaucher disease.

NEURONOPATHIC SUPPLEMENT

Zavesca Trial for Type 3 Gauchers Disease Halted

In 2004, Actelion took over responsibility for the miglustat study in type 3 Gaucher disease (GD3). This was an international effort involving two expert centres led by Dr Ashok Vellodi, London, UK and Dr Raphael Schiffmann, Bethesda, USA writes Dr Hélène Peyro-Saint-Paul, Global Brand Leader Zavesca, Actelion Pharmaceuticals:

'Thirty patients aged 2-20 years were enrolled by May 2004, and 19 were under 12. One patient on the study had previously had a bone marrow transplantation. In the first 12-month phase, 20 patients received a combination of their usual ERT regimen plus miglustat whereas 10 were maintained on ERT only as the control group. In the following 12 months all patients were on combination of ERT plus miglustat 200 mg three times a day. The maximum dose of miglustat was 200 mg tds; the dose was adjusted in children according to body surface area.

'Miglustat's wide tissue distribution including access to brain and lung was the rationale to evaluate if this drug could improve the neurological and pulmonary status of patients. Since GD3 is typically characterised by neurological manifestations, lung dysfunction, as well as organomegaly and blood abnormalities, our study was designed to look at all these key organs.

'On October 31st, the investigators and the Actelion's team met to review the 24-month results. The final consensus was to end the study as it could not demonstrate effects on the primary end-point of eye movements abnormalities.'

Investigator's Statement

Dr Ashok Vellodi, principle investigator for the study at Great Ormond Street said "We were very disappointed that the results did not show an effect as we had all hoped. Paediatric trials involve parents just as much as they do their children, and we fully appreciate how difficult this has been for the parents. It was particularly frustrating that the recruitment took so long and that this resulted in the trial taking nearly twice as long to complete as was originally planned. We would like to express our gratitude to all the families who participated in the study, and we hope that the results will help us, both in our understanding of this disease as well as in developing future therapies."

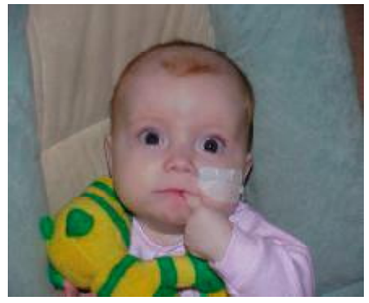
The Families

A parent of a child on the trial told the Gauchers Association: "The cessation of the trial is a bitter disappointment. Of course we understand that the purpose of the trial was to evaluate the effectiveness of a drug and as such there is always the risk it might not actually give sufficient evidence that it works. However my child has had to go through so much in the last three years. It has been very hard; the problems of persuading her to swallow the pill, the impact of the diarrhoea and the real challenge of having to complete all the tests.

We must now look to the future and hope that other options will soon become available."

Type 2 Gaucher Disease Research Project Announcement

Ellie Carter died on 9 February 2004 aged seven months. Ellie had Type 2 Gauchers 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. The ELF fund has raised £40,000 since Ellie died. The trustees of the Foundation have asked the Gauchers Association to administer the fund and identify a suitable research project to meet these aims.



Ellie Carter

The Gauchers Association would like to **invite research applications in the area of Type 2 Gauchers disease for consideration**. All applications must be made to the Gauchers Association in writing. The Medical Advisory Board of the Gauchers Association will review the applications. If you would like to submit a research application the Association please contact: Tanya Collin-Histed: ga@gaucher.org.uk or telephone: 00 44 (0) 1453 549231.

The Gauchers Association congratulate Jill and Ian Carter on the birth of their second son **Charlie** in October 2006. Charlie was born after Jill and Ian had Pre-Implantation Genetic Diagnosis (PGD). Their journey through PGD was followed by the BBC and was shown on BBC1 on Tuesday 14 November in the programme 'A Child Against All Odds' presented by **Prof Lord Robert Winston**.