

***Please circulate this update to all members of your organisation***

### ***Annual General Meeting 2007***

GOLD's Annual General Meeting will be held on Thursday 25<sup>th</sup> at 11.30 am, at the American Society of Human Genetics meeting in San Diego. Our Guest Speaker will be Dr William Gahl, who will speak on the subject "Advocacy for Rare Diseases"

Further details will be announced when the meeting notification is issued 3 months before the meeting. When you receive this, please return the

### ***GOLD Management Council***

Dr Ed Wraith and Mrs Christine Lavery have resigned from the Management Council. The Management Council thank them for all their work on behalf of GOLD. As both had a further year to serve, the Management Council have co-opted new members to the council to serve for the remaining period. We are very pleased to welcome to GOLD's management council:

**Professor Timothy Cox.** Tim, who delivered the Guest Lecture at GOLD's AGM last year is Professor of Medicine and Honorary Consultant Physician at Addenbrooke's Hospital, Cambridge UK.

**Dr Maurizio Scarpa.** Department of Pediatrics, University of Padova, Centre for Rare Disorders. Maurizio is co-founder of Brains for Brain, which is featured in this Update.

A call for nominations to the Management Council, with the retirement by rotation of 4 members of the council, will take place shortly.

### ***Sponsored Swim for GOLD.***

Thanks to everyone who sponsored me for this event. This raised just over £1,700 (US\$3350; €2,500) for GOLD, including Gift Aid, which enables tax to be reclaimed from UK taxpayers.

### ***Brains for Brain***

Brains for Brain is a newly founded European Task Force on Brain and Neurodegenerative Lysosomal Storage Diseases, led by Dr Maurizio Scarpa, University of Padova and Dr David Begley, King's College London. The initiative was conceived at a workshop in Madrid, March 2007 focusing on addressing current problems of overcoming the blood brain barrier to deliver therapy for LSDs.

The mission of Brains for Brain is to catalyse research in the field of neurodegeneration and lysosomal diseases. Brains for Brain is formed as an association of highly distinguished scientists and clinicians working together to create and coordinate effort towards the comprehension of the pathophysiology processes of neurological disorders occurring in lysosomal storage disorders. The implementation of knowledge of blood brain barrier function and the development of new molecular and or biochemical strategies to overcome the blood brain barrier and treat neurological disorders is a central part of the effort.

The achievement of these targets will allow the development of new approaches for therapy of the neurological complications of LSDs patients. For more information on Brains for Brain, see the GOLD website ([www.goldinfo.org/membernews.aspx](http://www.goldinfo.org/membernews.aspx)) or the Brains for Brain website ([www.brains4brain.eu](http://www.brains4brain.eu))

## ***Update on Requests for Proposals from GOLD member organisations.***

Requests for proposals from 3 member organisations were announced in the last Update, and posted on the GOLD website. The UK Gaucher Association, The National Tay Sachs and Allied Diseases Association and The Canadian MPS Society were extremely impressed with the quality of the research proposals they received. The awards made were:

**The UK Gaucher Association** awarded a grant to Professor Mia Horowitz, Dept Cell Research and Immunology, Tel Aviv University, Ramat Aviv, Israel. This is for a 3 years study, 15 hours a week.

Purpose of the project:

- To test the correlation between endo-H sensitivity in different cell types and neuronopathic Gaucher disease severity.
- To identify interactors of mutant glucocerebrosidase variants, that participate in its ERAD.
- To test whether ER stress caused to brain cells in patients with type 2 or 3 Gaucher disease leads to their death through apoptosis or autophagy

**The National Tay Sachs and Allied Diseases Association 2007 Research Initiative** awarded two grants. Recipients are:

1. Susan L. Cotman, Ph.D., Massachusetts General Hospital  
"Small molecule screening to identify modifiers of lysosomal trafficking, a putative therapy for Batten Disease"
2. Miguel Sena Esteves, Ph.D., Massachusetts General Hospital  
Douglas R. Martin, Ph.D., Auburn University.  
Gene Therapy for Tay-Sachs Consortium  
"AAV-mediated gene therapy for Tay-Sachs disease: Vector selection for pre-clinical development"

**The Canadian MPS Society** also awarded two grants for 2007:

3. \$40,000.00 grant to Dr. Lorne Clarke UBC, Vancouver for Biomarker Research
4. \$40,000.00 grant to Dr. Murray Potter, University of Western Ontario, London, Ontario for MPS II Blood Brain Barrier Research.

The full grant details are on the Canadian MPS Society's website - [www.mpssociety.ca](http://www.mpssociety.ca).

## ***Multidimensional Clinical Assessment Tool for Batten Disease***

Children and young people with Batten Disease, in the UK, are invited to participate in a project aiming to develop a clinical assessment schedule to monitor symptoms and disease progression. Participants will be asked to attend 4 six monthly assessments over a 2 year period either at the Evelina Children's Hospital in London or at a more convenient local centre or school. Assessments are expected to take between 60 and 90 minutes each. They will consist of a clinical neurological and developmental examination together with a short audio recording of speech in some cases.

If you are interested in participating please contact Dr Ruth Williams, Consultant Paediatric Neurologist, The Evelina Children's Hospital, for further details: telephone, 0207 188 4004; email, [ruth.williams@gstt.nhs.uk](mailto:ruth.williams@gstt.nhs.uk) or The Batten Disease Family Association at [bdfa.info@btinternet.com](mailto:bdfa.info@btinternet.com)

## **CLIMB Book**



CLIMB, Children Living with Inherited Metabolic Disease have published a book "Inherited Metabolic Diseases - a guide to 100 diseases" including LSDs, written by metabolic disease specialists and edited by Climb's Executive Director, Steve Hannigan.

The book is available, with a 5% discount from CLIMB  
<http://www.nicmd.org.uk/ecomcart/list.php>

## Gaucher Association 15th Anniversary Conference

The video of the Gauchers Association 15<sup>th</sup> Anniversary Conference is now available to view on the GOLD website. Presentations available include Scientific, clinical and family experiences. The Conference dinner featured speeches by Jeremy Manuel, chairman of the Gauchers Association, and the Guest Speaker, Sir Michael Rawlings, Chairman of the National Institute of Health & Clinical Excellence (NICE).

### **Presentations:**

Pathophysiology of nGD Dr Ashok Vellodi  
Auditory Involvement and Learning Dr Pauline Campbell  
Neurological Features and Complications Dr Catherine DeVille  
Blood Brain Barrier Dr David Begley  
Future Directions Dr Ashok Vellodi  
A Personal Story Laurena Simpkin  
A Dad's Story Abdul Waheed  
Future Trials and Therapies Prof. Ari Zimran,  
Bone Collaboration Project Prof. Timothy Cox and Dr. Patrick Deegan

Growing old with Gaucher Disease Dr Atul Mehta  
Alan Gordon Memorial Award Presentation Robert Gordon  
Doctors – Patient – Partnerships, how the past can address the future Prof. Timothy Cox,  
'Efficiency versus Equity: Should society fund orphan treatments?' Hannah Hyry,  
Specialist Commissioning for Gaucher disease and other LSDs Dr. Edmond Jessop,  
Living with Type 3 Gauchers disease Jo Bardoe  
Living with Type 1 Gauchers disease Daniel Long

A CD of the Anniversary Conference and after dinner speakers will be sent out with the next newsletter of the Gaucher Association.

## Upcoming Events

For further details, please see the Calendar of Events page on the GOLD website. If you are organising a meeting, which is not yet listed, and would like to have it listed on GOLD's website or in the next Update, please contact [enquiries@goldinfo.org](mailto:enquiries@goldinfo.org).

### **July 12-15th, Rochester, NY**

Batten Disease Support and Research Association (BDRSA) Annual Meeting.

### **July 14 - 17th, Rochester, NY**

11th International Congress on Neuronal Ceroid Lipofuscinosis (Batten Disease)

### **July 26 - 28th, Ann Arbor, MI, USA**

International Society for Mannosidosis and Related Diseases.  
2nd International Symposium for Glycoprotein Storage Diseases.

### **August 16 - 19th, Milwaukee, WI, USA**

National Niemann Pick Disease Foundation  
15th Annual Family Support and Medical Conference.

### **September 4-7th, Hamburg, Germany**

44th Annual Symposium: Society for the Study of Inborn Errors of Metabolism (SSIEM)  
International meeting, open registration

### **September 27 - 30th, Perugia, Italy**

European Study Group for Lysosomal Diseases  
16th ESGLD Workshop.

### **October 5 - 6th, Aranjuez(Madrid) Spain**

Spanish MPS Society.  
3rd National Congress

**October 18th, 7-11pm, Tribeca Rooftop, New York, NY, USA**

National Tay Sachs and Allied Diseases Association  
50th Anniversary Gala

**October 23 - 27th, San Diego, California, USA**

American Society of Human Genetics **October 25th, 11.30 - 1pm San Diego, California, USA**

GOLD Annual General meeting. Guest Lecturer, William A Gahl MD PhD.

**October 25th, 1-2pm San Diego, California, USA**

Pharmacological Chaperones: A Novel Investigatory Approach for Treating a Broad Range of Genetic Diseases.

Lunchtime Symposium sponsored by Amicus Therapeutics.

**Contact GOLD:**

If you have any comments or suggestions, please send them to:

Ann Hale: [ann.hale@goldinfo.org](mailto:ann.hale@goldinfo.org),

E-mail: [enquiries@goldinfo.org](mailto:enquiries@goldinfo.org)

fax/phone +44 (0) 1494 870708

Mail: Global Organisation for Lysosomal Diseases  
PO Box 609  
Chalfont St Giles  
HP8 4WU  
UK

GOLD forums: <http://www.goldinfo.org/forums/Login.aspx>