

Please circulate this update to all members of your organisation

Happy New Year

On behalf of GOLD's Management Council, I would like to wish all our members a very happy New Year, and hope you had an enjoyable Holiday Season.

Annual General Meeting 2007

GOLD's Annual General Meeting was held on Thursday 25th October at the American Society of Human Genetics meeting in San Diego. The Annual Report and accounts were issued to all member organisations, and are available on the GOLD website



Our Guest Speaker was Dr William Gahl, MD PhD Clinical Director, National Human Genome Research Institute, who spoke eloquently and movingly on the subject "Advocacy for Rare Diseases." He described the challenges and frustrations of new therapeutic interventions in extremely rare "ultra orphan" diseases.

To access Dr Gahl's guest lecture:

- Go to the GOLD website www.goldinfo.org
- Select > Education and Information > Video Presentations
- Log in with your user name and password*
- When the Video Presenter loads, select the GOLD tab in the right hand column.
- Select the title "Advocacy for Rare Diseases".
- Select "Play" in either windows media or real player.
- The presentation will load.

Dr Gahl kindly allowed us to film his Guest Lecture, which is now available on the GOLD website under the Member area video presentations.

*If you are not yet registered as a member user of the website:
Go to www.goldinfo.org/forums/register.aspx
Enter your email address as user name and choose a password.
Select the organisation of which you are a member.
You can then access the Video presentations.

Next Annual General Meeting - 2008

The GOLD AGM in 2008 will be held at the conference which will be attended by the majority of our member organisations. To suggest a suitable conference, please contact Ann Hale or a member of the Management Council.

GOLD Management Council

Dr Michael Beck, Dr Greg Grabowski, Professor John Hopwood and Dr Bill Sly all retired by rotation from the management council. Dr Bill Sly did not stand for re-election. GOLD thanks Bill for all the work he has done on our behalf, since he was one of the founding members of GOLD. Bill assures us he remains a staunch

supporter and advocate for GOLD, and has agreed to be our Guest Lecturer at the next AGM.

The nominees for the Management Council were: Dr Michael Beck, Dr Greg Grabowski, Professor John Hopwood and Professor Mark Sands. As four places were vacant, all were deemed elected.

Canadian MPS Society - Research Grant Request for Applications

The Canadian MPS Society has issued a Research Grant Request for Applications. The grant is for \$40,000.00 (CND) for research into mucopolysaccharide diseases. The application deadline is Feb. 14, 2008 with a funding date of March 31, 2008. Application forms may be obtained from Kirsten Harkins at the Canadian MPS Society (kirsten.mpssociety.ca@telus.net) or downloaded from the GOLD website at <http://www.goldinfo.org/membernews.aspx>. Applications must be made direct to the Canadian MPS Society – full details are on the RFA form.

10th International MPS Symposium – Call for Abstracts

The Scientific Committee for the 10th International Symposium on Mucopolysaccharide and Related Diseases invites the submission of abstracts describing all aspects related to MPS.

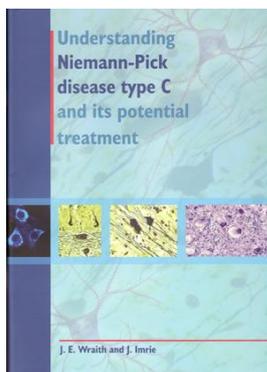
Abstracts should be submitted within the following categories:

- I. Disease Pathophysiology: The skeleton and connective tissue in the MPSs.
- II. Disease Pathophysiology: The CNS and disease:
- III. Disease pathogenesis and animal models:
- IV. Therapies for the MPSs
- V. Neonatal screening for the LSDs

Presentation submissions will be reviewed for presentation as either an oral or poster presentation. The assignment of presentation type is at the discretion of the scientific committee and will be confirmed upon acceptance.

Abstracts may be submitted at www.mpssymposium2008.com.

NPDG-UK Niemann Pick C Book



The Niemann Pick Disease Group, UK (NPDG-UK) has published a book, ***Understanding Niemann-Pick disease type C and its potential treatment***, in association with the Italian and German Niemann Pick support groups.

The book, written by Ed Wraith and Jackie Imrie, is a comprehensive review of NP-C, covering the history of the disease, genetics and inheritance patterns, epidemiology, pathophysiology and neuropathology, clinical manifestations and diagnostic strategies, treatment strategies and patient & family support.

This excellent and clearly written reference book is available free of charge from the NPDG-UK. Contact Jackie Imrie, Niemann-Pick Clinical Nurse Specialist. (Jackie.Imrie@CMMC.nhs.uk)

National Gaucher Foundation launches Gaucher Kids website

The National Gaucher Foundation (NGF) has developed a new website dedicated to kids and young adults with Gaucher or any other LSD. It will allow children and young adults from 6 to college age to enjoy the freedom of having a place to call their own so that they can communicate with others who have like interests and issues in common. The new site is at www.gaucherkids.org

Expressions of Hope Exhibition

"Expressions of Hope" is a new exhibition at the National Museum of Health and medicine (NMHM.) The exhibition, sponsored by the National Gaucher Foundation and Genzyme Therapeutics, showcases 32 pieces of art by people affected by an LSD. It was created to express the courage of the global LSD community and to generate awareness and understanding of people living with LSD. Patient organisations around the world encouraged their members to share their experiences and emotions through all types of art, to symbolise the feelings of hope whilst exploring the realities of living with one of these conditions.

The NMHM exhibition opened on November 8th 2007, and features an additional three pieces of artwork by Californian artist Ted Meyer, who also has Gaucher Disease.



Tina Johnson Bowen, Mother of Grant Bowen, artist on behalf of Gaucher Disease.

At the opening of the Expressions of Hope Exhibition, NMHM, Nov 8th 2007.

Photo credit:
Steven Kruger Senior
Staff Photographer,
NMHM.

The exhibition runs until March 2nd 2008. If you are in the Washington area, this exhibition, described by the Washington Post as "touching and inspiring" is well worth a visit. Admission and parking are free and the museum is open every day from 10.00am to 5.30pm. The museum is located on the campus of the Walter Reed Army Medical centre, Silver Spring, Washington DC, reached via the main entrance at Georgia Avenue and Elder St. Adults will be asked to present photo ID. For further details, view www.nmhm.washingtondc.museum

Batten Disease Training Days

The Batten Disease Family Association, UK and the Batten Disease Professional Interest group held a very successful Training Day at Guy's Hospital, London in November 2007. Around 30 professionals from a range of backgrounds participated, whose feedback confirmed they went away with increased, relevant knowledge about Batten Disease and the resources available and useful networking opportunities.

Following the success of the course, a second opportunity to participate will be available on Thursday 17th April 2008. There will be a morning program of short lectures followed by afternoon workshops. For more information, see the Members' News page on the GOLD website, <http://www.goldinfo.org/membernews.aspx>. Or 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 or The Batten Disease Family Association at bdfa.info@btinternet.com

Multidimensional Clinical Assessment Tool for Batten Disease

This Clinical assessment project is currently recruiting. 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.

To enquire about participation, contact Dr Ruth Williams, Consultant Paediatric Neurologist, The Evelina Children's Hospital: telephone, 0207 188 4004; email, ruth.williams@gstt.nhs.uk.

Upcoming Events

FEBRUARY 2008

February 13-15, Las Vegas, Nevada, USA

WORLD Symposium
Lysosomal Disease Network

MARCH 2008

March 1-2nd, Plovdiv, Bulgaria

3rd Eastern European Conference on Rare Diseases & Orphan Drugs:
"Rare Diseases - Prevention, Diagnosis, Treatment"

March 27 - 30th Tampa, FL, USA

30th Annual Family Conference.
National Tay Sachs and Allied Diseases Association

APRIL 2008

17 April London UK

Batten Disease Training Day
Batten Disease Professional Interest Group and Batten Disease Family Association

MAY 2008

May 29 - 31st, Hyatt Regency Sacramento, California

3rd Lysosomal Diseases and the Brain Conference.
Children's Gaucher Research Fund

JUNE 2008

June 4-7th, Budapest, Hungary

8th meeting of the European Working Group for Gauchers Disease

June 26-29th 2008, Vancouver, BC, Canada

10th International Symposium on MPS and Related Diseases. Hosted by Canadian MPS Society.

June 28-29th 2008, TBA

Niemann Pick Disease Group UK Annual Family Conference

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

Contact GOLD:

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

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