

SSIEM Newsletter February 1997

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35th ANNUAL SYMPOSIUM, Gothenburg, Sweden, 2nd-5th September 1997

The arrangements for this meeting are now well advanced, and you will see from the second notice included with this mailing, that we have an excellent scientific programme. As with last year, all abstracts that are submitted in time and accepted for the symposium will be published in a JIMD supplement to be published before September. The closing date for abstracts is the 1st May and for registration the 31st May. Register early to avoid disappointment!

34th ANNUAL SYMPOSIUM Cardiff, 10th-13th Sept. 1996

By Graham Shortland

The 34th Annual Symposium was well supported by members and non members of the Society alike. This in fact reached the point prior to the Symposium when some registrations had indeed to go on a waiting list because of the high number of registrants. Altogether 425 active delegates attended the Symposium with large numbers able to also attend the full and active social programme throughout the Symposium. A small, but nevertheless important contingent of social delegates had a very enjoyable conference travelling as far a field as the Wye Valley and Bath in the County of Avon and local museums.

All the scientific sessions were well attended and appeared to be appreciated and enjoyed with a high standard of presentation. This included the quite outstanding Komrower lecture "G protein-coupled signal transduction pathways in human disease" by Dr. Spiegel. Once again poster walks appeared to be very popular and well appreciated.

The Local Organising Committee are now recovering from the rigors of the conference, and in particular we are very pleased that delegates did not require the use of their Symposium umbrella with fine weather prevailing throughout the conference. Financially the conference was also a success and made a small surplus to be carried forward to Gothenburg.

Secretary's Column

Welcome to the February newsletter. This year promises to be another interesting one for those involved in biochemical genetics. We have two major symposia to attend, the first the [International meeting in Vienna](#) in May and then our own [symposium in Gothenburg in September](#). The second notice, registration form and abstract form for the Gothenburg meeting are with this mailing. The interest in inherited metabolic disease is also reflected in the increasing number of submitted papers to our journal, the JIMD. In addition there is the lively internet correspondence that is generated by [Metab-L](#) and [Inborn-errors](#), two e-mail based discussion groups. Those of us who subscribe to these will have noticed that there are a number of individuals who are profligate contributors. Others, I expect, are more voyeurish in their approach. These mailing lists are free to subscribe to and encourage the dissemination of information and the sharing of experience. If you have a computer and a modem there is no reason not to join. Our own Web page, as featured in the last newsletter,

is developing well and now contains the text of this newsletter. In addition it gives information about the SSIEM, the contents of each issue of the JIMD, and has links to other relevant web pages such as the OMIN database. Please do visit it and, having done so, provide feedback in the relevant section of the page. Another exciting development, also linked to the computer era, is the release in March, of a version of The Metabolic and Molecular Basis of Inherited Disease on compact disc. I am told by Charles Scriver that this edition, called version 7.5, has updated information for approximately half the book plus new chapters on cancer genetics. The additional and updated material is equivalent to more than 600 pages. As with the 7th edition [Blackwell Science](#) are offering SSIEM members a 15% discount of the list price. The March issue of the American Journal of Genetics will contain an article on Professor Scriver's view of the state of the molecular basis of inherited disease in the 1990s. The title of the article is Realities and Virtual Realities of Inborn Errors of Metabolism: Biochemical Genetics in the Molecular Genetic Era.

Last year's symposium was very successful, both in terms of attendance, and content. The lectures and free communications were excellent and the quality and diversity of the poster presentations reflects the strength and depth of the society. The provision of a JIMD supplement containing abstracts, which can now be easily referenced, has been well received. Despite some early misgivings the production of this supplement went ahead very smoothly. Meeting old and new friends and colleagues and discussing ideas is a major stimulus to our work and the importance of such scientific and social exchange should not be underestimated. Each year's symposium requires a huge amount of planning and by far the greatest share of this falls to the local organising committee. We are, consequently, very grateful to Dr Graham Shortland and all his team for their work. They can now relax and bask in the glory of a successful conference.

Report from Council

Since the previous newsletter corresponding members and council met on 11th Sept and council on 29th November 1996 in Sheffield. We welcome Professor Kim Bartlett onto council. The current members of council are as follows:

- i Dr Brian Fowler (Chairman)
- i Dr John Walter (Hon Sec)
- i Dr Jim Bonham (Hon Treasurer)
- i Dr Rodney Pollitt (Editor-in-Chief, JIMD)
- i Dr Garry Brown
- i Dr Peter Clayton
- i Prof Wolf Endres
- i Dr Jean Kirk
- i Dr Albert van Gennip
- i Professor Kim Bartlett

Corresponding members are as follows:

- i Mr U Caruso, Italy
- i Dr W Sperl, Austria
- i Dr H Ogier, France
- i Dr K M Gibson, USA
- i Prof J Jaeken, Belgium
- i Dr C Jakobs, Netherlands
- i Dr V Kozich, Czech Rep
- i Dr E A Kvittingen, Norway
- i Prof C Scriver, Canada
- i Prof Y. Eto, Japan
- i Prof M Ugarte, Spain
- i Prof K Ullrich, Germany
- i Dr B Wilcken, Australia

For a list of the new members: 1996 [New Member List](#)

Please contact your local corresponding member to express a view on any aspect of the Society. The full addresses of Council and Corresponding members are in your members' handbook.

Material for the newsletter

This newsletter is sent in early February and August. Please send any material for inclusion, at least one month before these dates, to

Dr J H Walter,
Honorary Secretary,
Willink Biochemical Genetics Unit,
Royal Manchester Children's Hospital,
Pendlebury,
Manchester
M27 4HA.
Fax: + 44 161 727 2137,
E-mail: John@willink.demon.co.uk

Training in metabolic disease - important! Under the current proposals, the EU will not recognise metabolic medicine as a subspecialty of adult medicine, paediatrics or clinical genetics. As a result there is a risk that those in training may be reluctant to enter this field with obvious long term consequences. It is important that there should be a forum to collect details of national training programmes and represent our interests.

At the SSIEM meeting in Cardiff a request was made for information but there has been little response from individual members. Please would you send details of training programmes or the names of those who should be contacted to :

Prof JV Leonard,
Metabolic Unit,
Institute of Child Health,
30 Guilford Street,
London
WC1N 1EH,
Fax +44 171 813 8496,
email: j.leonard@ich.ucl.ac.uk

or to
Dr J H Walter,
Willink Biochemical Genetics Unit,
Royal Manchester Children's Hospital,
Pendlebury,
Manchester,
M27 4HA
Fax +44 161 727 2137,
email: john@willink.demon.co.uk

International Dietitians Meeting at the SSIEM

By Anita McDonald

Although dietary management is an integral part of treatment for so many inherited metabolic diseases, few dietitians have traditionally attended the SSIEM International Meeting. This is because dietitians consider

this to be a more scientific than clinically orientated meeting, although in reality there is always much new information about disorders and therapies and it is invaluable for dietitians with an interest in metabolic disease. Therefore in 1996 we held the first international metabolic dietitian session at the SSIEM in Cardiff 1996. This was not only to attract dietitians to attend this meeting, but we saw it as an ideal opportunity for dietitians working in metabolic disease to discuss our own original research work.

We were allocated one afternoon session and the topic chosen was "Dilemmas of Protein Requirements in Metabolic Disease". We planned one keynote speaker on protein requirements, original research presentations, a round-table discussion, with our own poster session. SHS kindly agreed to publish our abstracts and offered a prize for the "best" published presentation or abstract. Despite these ambitions, we only anticipated a small attendance, perhaps with a handful of dietitians. In fact, we could not have been more wrong; the session prove more popular than we could have imagined. On the day we had an attendance of at least 70 delegates and 50% of these were dietitians. An excellent overview on "Protein Quality and Requirements" was given by Professor Millward followed by three original research presentations by dietitians. All papers were stimulating and thought provoking, discussion was fast and furious and this session perhaps created more questions than it answered. However, all delegates went home very satisfied and the post conference feedback has very clearly indicated that not only should this meeting be repeated but even extended!

Several entries were received for the dietetic SHS award, and the winner was Marjorie Dixon from Great Ormond Street, London on "Growth in Disorders of Propionate Metabolism". SHS have kindly agreed to publish a compilation of papers from this session and copies will be available in September 1997. What about the future? The SSIEM Council have kindly agreed that we can hold a further dietitians' meeting at the SSIEM in Gothenburg in 1997. This will be on Thursday 4th September. Subject matter will include "Dietary Management of Galactosaemia" and "Dietary Management of Tyrosinaemia (Types I, II and III)". We are planning two keynote speakers and original research presentations, as well as a dietetic poster session. Dietitians already on our mailing list will receive further information about this session as well as abstract forms. Any member of the SSIEM wishing to present a paper in this session should contact the meeting co-ordinator (name and address below) directly. Further details will be published in the Gothenburg meeting notices. We would like to invite all delegates interested in dietary management to our session.

Anita MacDonald,
Dietetic Department,
Birmingham Children's Hospital,
Ladywood Middleway,
Ladywood
Birmingham
B16 8ET

Parent Groups

13th Parent's Conference of the Association for Glycogen Storage Disease (UK) to be held in London from 24th - 25th May 1997. For further information please contact::

Mrs. Ann Phillips,
Association for Glycogen Storage Disease (UK),
9 Lindop Road,
Hale,
Altrincham,
Cheshire
WA15 9DZ,
UK

Link to Association for Glycogen Storage Disease Homepage :

<http://www.compulink.co.uk/~embra/agsdhome.html>

15th Annual Conference of The Society for Mucopolysaccharide Diseases

Stakis Country Court Hotel Northampton, 12th - 14th September 1997.

For further information please contact:-

The MPS Society,
55 Hill Avenue,
Amersham,
Bucks
HP6 5BX,
UK

Link to MPS Society Homepage : <http://www.vois.org.uk/mps/>

16th Annual Conference of the Research Trust for Metabolic Diseases in Children

13th - 14th September 1997 at the Stakis Grand Hotel, Hanley, Stoke-on-Trent, UK.

For further information please contact:-

Research Trust for Metabolic Diseases in Children,
Golden Gates Lodge,
Weston Road,
Crewe,
Cheshire
CW1 1XN

GHB Parent Support Group (from Dr. K. Michael Gibson)

At present, the approximate number of patients detected with succinic semialdehyde dehydrogenase deficiency (4-Hydroxybutyric aciduria) is approximately 100-150. The patient population thus far studied at Baylor University Medical Center in Dallas has originated from essentially every region of the globe, although to our knowledge we have yet to identify a patient living in Africa or South America. As expected for an autosomal recessive disorder, the phenotypic presentation demonstrates substantial variability, from very mildly affected patients to those whose clinical presentation is quite severe. Over the years, we have had requests from a number of families for names and addresses of other families with affected children. The objective is to provide emotional, moral and other forms of support and dialogue between parents. For this reason, we have decided to begin a support group for these patients and family members, named the GHB (for gamma-hydroxybutyric acid, or 4-hydroxybutyric acid) Parent Support Group.

If you, or a colleague, are caring for a patient(s) with 4-hydroxybutyric aciduria, you may wish to share this information with that family. Interested families are requested to supply names of parents and patients, mailing addresses, fax, phone and e-mail address if available. All information will be maintained in the strictest confidence. An address book of families will be developed and circulated only within the circle of families listed.

If you are interested, please forward the above information to:

Dr. K. Michael Gibson,
Institute of Metabolic Disease,
3812 Elm Street,
Dallas,
TX,
USA;
phone 214-820-4749;
fax 214-820-4853;
e-mail km.gibson@baylordallas.edu.

Meetings & announcements

The 11th Annual Meeting of the Australasian Society for Inborn Errors of Metabolism (ASIAM)

Will be held in Perth on July 25th and 26th 1997.

This year's guest speakers are Professor Bob Desnick and Professor James Leonard. The meeting is held in conjunction with the annual scientific meeting of the Human Genetics Society of Australasia. Also meeting in Perth that same week, from the 21st to 25th July is the International Union of Anthropological and Ethnological Sciences and the Society for Human Biology. For these latter meetings there is a large line-up of invited speakers, which includes Peter Harper, Bob Williamson, Malcolm Ferguson-Smith, and several others.

This might be an excellent excuse to visit Australia. Further details of the ASIEM meeting could be obtained from either Dr Bridget Wilcken, or Dr. Judith Hammond, the Honorary Secretary. The address for both of us is-

Department of Biochemical Genetics,
The New England Children's Hospital
P. O. Box 3515
Parramatta 2124
NSW
Australia.
Fax +61 2 9845 3121
Email: Bridgetw@nch.edu.au

11th International Symposium Chemistry and Biochemistry of Pteridines and Folates

June 15-20, 1997

January 15, 1997: Last day for early registration fee and for abstracts to be considered for oral presentations,

February 28, 1997: last day for abstracts for poster sessions

There is information available also on the internet at <http://www.kfunigraz.ac.at/imcwww/b-gaden97.html>

[A summary of the symposium and daily list of the lectures are given here.](#)

[Information on the journey to the symposium .](#)

Further information from:

Dr. Hartmut Rokos,
Robert-Koch Institute,
FB1,
Berchtesgarden/Germany,
Nordufer 20,
D-13353 Berlin/Germany.
Fax: +49 30 4547 2605,
e-mail: RokosH@RKI.de.

Molecular Medicine and Inherited Diseases: Retrospections and Outlook

This symposium in honour of Prof. Richard Gitzelmann will be held at the University Children's Hospital in Zurich, Switzerland, on April 17, 1997, beginning at 2.00 p.m. Speakers will include F. H. Sennhauser, C.C. Kuenzle, C. Bachmann, V. A. McKusick, J. Spranger, C. Scriver, Dr. Valle, H. Galjaard and G. Duc. Topics will include advances in human, medical and biochemical genetics over the past forty years, novel results in human morphogenesis, future perspectives for genetic medicine, and social implications of advances in human genetics. Admission is free.

For more information please contact

Mrs. Irene Cathomen (FAX +41-1-266-7167, Phone +41-1-266-7310),

B. Steinmann or A. Superti-Furga at the Division of Metabolic and Molecular Diseases, University Children's Hospital, Zurich, Switzerland.

4th International Symposium on Brain Dysfunction - Neurogenetic Disease:

From Molecule to Patient + International Prize on Brain Dysfunction Research, Troina, Sicily (Italy), September 24-26, 1997. Sponsored by the Oasi Institute for Research on Mental Retardation and Brain Ageing

Various aspects of neurogenetic diseases: phenotype, pathology, pharmacology, biochemistry, genetics, animal models, and treatment will be discussed. We hope that this international meeting will further promote research in this field in order to understand pathogenesis as well as to develop new therapeutic approaches to genetic diseases affecting the nervous system.

Three lectures per session will be presented by invited speakers. The last hour of each session will be devoted to free communication by other participants at the Symposium. The afternoon session on the second day of the Symposium will be devoted to the presentation, by the author, of the paper awarded the Oasi International Prize for Brain Dysfunction Research. Deadline for the presentation of abstracts for free communications is July 15th, 1997.

The Oasi Institute invites the submission of original, unpublished research papers to compete for the 1997 award on the following subject: Neurogenetic Disease: From Molecule to Patient The Award: (a) US\$ 10,000; (b) invitation, with all expenses paid, to present the award-winning paper at the 4th International Symposium on Brain Dysfunction in Troina, Sicily (Italy) in September 1997; (c) publication of the manuscript in the journal *Developmental Brain Dysfunction* Deadline for the submission of manuscripts is April 30th, 1997. The rules for submission can be obtained from the Organising Secretariat. More information is available on the Internet: <http://www.oasi.en.it/prize.htm>

Symposium Scientific and Organising Committee:
R. Ferri, Troina, Y. Suzuki, Tokyo, J. R. Villablanca,
Los Angeles, CA.

Organising Secretariat:
Mrs. H. Cerro,
4th International Symposium on Brain Dysfunction,
Oasi Institute for Research on Mental Retardation and Brain Ageing,
I-94018 Troina (Italy).
Tel. ++39-935-93611,
Fax ++39-935-653327,
e.mail hcerro@oasi.en.it

The Third International Workshop on Resistance to Thyroid Hormone

Aspen, Colorado, USA, October 12-14 1997, Given Institute and The Mountain Chalet in Aspen, Colorado To be held prior to the 1997 Annual Meeting of the American Thyroid Association in Colorado Springs. The Workshop will focus on clinical and basic aspects of RTH, mechanism of thyroid hormone action and animal models of RTH with brief presentations with ample time for informal discussion and posters. Particular emphasis will be placed on participation by successful young investigators, junior faculty and minorities. Land transportation from Aspen to Colorado Springs will be available on the morning of Tuesday, October 14.

For information, contact:
The Third International Workshop on Thyroid Hormone Resistance,
c/o Dr. Samuel Refetoff,
The University of Chicago (MC3090),
5841 South Maryland Avenue,
Chicago,
IL 60637.
Tel (773) 702 6939,
Fax (773) 702-6940.

The Amsterdam International Medical Summer School

Molecular and genetic aspects of Endocrine and Metabolic Diseases, 21st July to 8th August 1997.

For further information please contact:-

The Amsterdam Summer University,

P. O. Box 53066,

1007 RB Amsterdam,

The Netherlands,

Tel: 31 20 6200 225,

Fax: 31 20 6249 368

8th Annual BIMDG Symposium, Institute of Child Health, London, Wed May 7th 1997.

Peroxisomal disorders.

Further information from

Dr PT Clayton,

Biochemistry Unit,

Institute of Child Health,

London

WC1N 1EH.

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