
SSIEM Newsletter

Number 29 February 1999

Secretary's column

This is the penultimate newsletter of the millennium. As we get nearer to the year 2000, as with many other organisations, we have to assess whether we are 2000 compliant. The members database is held in Microsoft access version 2. A visit to Microsoft's millennium web page states that this software isn't compliant. You will understand then why you might receive a demand for subscriptions payments from 1900 to the present day. This will obviously be very good for the SSIEM's financial standing! Fear not though I am sure we will have sorted out this problem.

Last year we had our most popular annual symposium ever. This year we look forward to meeting in Genoa. Ubaldo Caruso and Roberto Cerone are leading the local organisers and have been working to ensure another successful meeting

Report from Council

Council and corresponding members met for their yearly meeting in York last September and there has been a further council meeting on 4th December 1998. At both meetings there has been discussion as to how abstracts should be selected for the symposium. The policy agreed by Council is discussed in detail in this newsletter.

We have also made considerable progress with the development of recommendations for training in metabolic disease. As the society becomes larger and more international we need to continue the debate as to what extent we wish to expand our role beyond the organisation of meetings. Please let us have your views on this or other matters.

Council members

Kim Bartlett

Jim Bonham (Treasurer)

Jane Collins

Brian Fowler (Chairman)

Jean Kirk

John Walter (Secretary)

Peter Clayton

Albert Van Gennip

Kurt Ullrich

Rodney Pollitt (Editor JIMD)

Corresponding members

Mr U Caruso, Italy

Dr W Sperl, Austria

Dr H Ogier, France

Dr K M Gibson, USA

Prof J Jaeken, Belgium

Dr C Jakobs, Netherlands

Dr V Kozich, Czech Rep

Dr E A Kvittingen, Norway

Prof C Scriver, Emeritus Corresponding Member

Prof Y. Eto, Japan

Prof M Ugarte, Spain

Dr S Schweitzer, Germany

Dr B Wilcken, Australia

Dr O Elpeleg, Israel

Prof R Giugliani, Brazil

Dr B H Robinson, Canada

Please contact your local corresponding member to express a view on any aspect of the Society.

Material for the newsletter should be to me at:

Willink Biochemical Genetics Unit,
Royal Manchester Children's Hospital
Manchester
M27 4HA

Fax: + 44 161 727 2137

email: John@jhwalter.demon.co.uk

Dr J H Walter

Honorary Secretary

July 1998

36th Annual Symposium at York

This meeting was the largest annual symposium to date with 621 delegates. It was based at the campus of York University just on the outskirts of the historic city of York. The facilities of the University were readily able to accommodate the conference, but the social programme struggled at times, the organisers having originally planned events that would accommodate a maximum of 450 people. The high point of the social programme was the symposium dinner in the magnificent surroundings of the National Railway Museum. Delegates were entertained before dinner by a traditional Yorkshire brass band and then dined on a platform between the carriages of steam trains.

The scientific programme started with a very popular pre-meeting 'workshop' on current advances in the use of tandem mass spectrometry for neonatal screening. The conference began in earnest with sessions on neurotransmitters and cerebral organic acidopathies. These were followed by presentations on defects of fat oxidation. The programme was completed by sessions on DNA technology and progress in understanding neurological diseases.

168 posters were selected for display from the 331 abstracts that were submitted, 6 were chosen for oral presentation. A further 130 abstracts were included in the abstract booklet. A new feature that was piloted at York were chaired poster talks, each presentation being 5 minutes long. Whilst these were very successful, some delegates have since expressed the view that they missed the attended poster sessions of previous meetings. It is clearly a problem that as the meetings get bigger it becomes harder to meet all the people that you would like to talk to.

The organisation was a collaboration by colleagues from two centres, Leeds and Sheffield, and was a tribute to the way that we were all able to co-operate. I would like to thank all those who helped to make this such a useful and enjoyable conference, organisers, delegates and commercial sponsors alike. I have to admit that I was previously unaware of the tremendous amount of work that goes into organising these conferences. I will certainly be more appreciative in the future!

Mick Henderson, on behalf of the local organising committee.

Selection of Abstracts

In the past it was possible to accept as posters, almost all submitted abstracts, that were not selected as oral presentations. The increasing popularity of the annual symposia has seen a concomitant rise in the number of abstracts submitted. Space restriction now dictates that there must be some limitation in the number of poster presentations. Furthermore some delegates have expressed an opinion that too many posters are detrimental to the overall quality of the symposia.

Abstracts are now published in a special JIMD supplement which is available prior to the symposia. This has many advantages, particularly that abstracts are now available for citation. However, due to the short time available between the closing date for submission and sending proofs to the publishers, it has led to more difficulties with judging abstracts.

This matter has been debated by council and corresponding members. It is accepted that a proportion of abstracts will need to be rejected on the grounds of quality and that not all can be accepted for poster presentations. Whereas in the past the judging of abstracts has been left primarily to local organisers a more robust and transparent method now seems advisable. The following principals have been agreed by council:

- | It is likely that the total number of poster presentations will be in the region of 200, but this may vary according to the nature of the meeting and space available.
- | The responsibility for judging abstracts will be the that of the SSIEM Council.
- | Council will appoint a panel of judges, one of whom will usually include a member of the local organising committee.
- | Abstracts will be either accepted for oral presentation, poster presentation or for publication in the JIMD supplement only.
- | The basis of acceptance and categorisation will be made wholly on the basis of scientific merit.

It is recognised that rejection, or acceptance for publication only, may create difficulties for individual members as institutions may not provide expenses for delegates who are not presenting posters. However it will be possible to provide letters of invitation if this will help and the SSIEM does provide a number travelling scholarships each year.

Although there will always be subjective elements in judging abstracts, council firmly believe that the only justifiable method of assessment is on quality alone. To this end we will also be looking at the feasibility of the judging being undertaken on abstracts rendered anonymous.

JH Walter

Meetings diary

Future SSIEM symposia 1999 to 2002

- | 1999 Genoa (7th-10th Sept)
- | 2000 Cambridge (13th-17th Sept)
- | 2001 Prague
- | 2002 Dublin

6th International Congress on Amino Acids.

Bonn, Germany, August 3-7th 1999. Molecular Medicine, Biochemistry, Nutrition, Pharmaceuticals. Abstracts will be published in the journal AMINO ACIDS, published by Springer and listed in Current Contents Life Sciences. The deadline for the submission of the titles is January 31st, 1999. The deadline for submission of abstracts with or without full manuscripts, for registration and payment of the congress fee is March 31st, 1999. Early registration is recommended due to a limitation to 600 participants. The registration fee is due latest by March 31st, 1999. For further information please contact:-

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Rheinische Friedrich-Wilhelms-Universität
Experimentelle Radiologie und Strahlenbiologie
Sigmund-Freud-Strasse 25
D-53105 Bonn, Germany
Fax: +49-228-287-4457
e-mail: hlink@mail.meb.uni-bonn.de*

4th Meeting of the International Society for Neonatal Screening

Stockholm, Sweden, June 13 - 16 1999. Scientific Program: Neonatal Screening- The sample and the sampling; New analytical techniques; Evolving programs; New treatment strategies; Neonatal thyroid screening and iodine deficiency disease; Neonatal screening for disorders such as hearing defects; Ethical, legal and health economical aspects; Biobanks; Neonatal screening in the new millennium. For scientific information, contact:

*Professor Agne Larsson
by Fax at +46-8-585-874-40 or e-mail to: agne.larsson@klinvet.ki.se
Conference Secretariat may be contacted :
ISNS 1999,
c/o Stockholm Convention Bureau,
P.O. Box 6911,
S-102 39 Stockholm,
Sweden.*

Home page: <http://www.stocon.se/isns>

The 13th U.S. National Newborn Screening Symposium

September 8-10, 1999, in St. Louis, Missouri.

*Contact James Baumgartner,
State Public Health Laboratory,
307 W. McCaarty,
Jefferson City,
Missouri 65102,
USA.
Fax to 573-522-8155 or e-mail baumgj@mail.health.state.mo.us*

First National Workshop On The Fish-Odour Syndrome And Dysfunctional Flavin Monooxygenases,

March 29/30 1999, National Institutes of Health, Bethesda, MD. USA.

The US Office of Rare Diseases of the National Institute of Health (Bethesda, USA) has uniquely joined forces with the Wellcome Trust (London, UK) to co-sponsor the First International Workshop on the Fish-Odour Syndrome and Dysfunction of the Flavin Monooxygenase System. This is the first official recognition, by both host institutions, that an enzyme of oxidative drug metabolism can be the underlying cause of a distressing inherited human disease. The Workshop will be held on the N.I.H. campus and registration is free. Accommodation will be available at nearby hotels at a reduced rate. For further information contact:

Dr. Steve Mitchell
Molecular Toxicology,
Division of Biomedical Sciences,
Imperial College School of Medicine,
Norfolk Place,
London
W2 1PG,
UK.
Fax: (44-171) 723 7535 or e-mail: s.c.mitchell@ic.ac.uk

The 1999 annual meeting of the Society for Inherited Metabolic Disorders

Lake Lanier Islands Hilton Resorts in Lake Lanier Islands, Georgia March 12-15, 1999.

Lake Lanier Islands is located 45 minutes north of downtown Atlanta. This year's program will include a symposium on inborn errors of membrane transport proteins, an area where exciting advances have been done in the past few years, both at their basic science and their clinical applications. As treatment improves, more of our patients reach adulthood, and also IEM have increasingly been recognised in adult patients; some of these issues will be discussed in another symposium. A workshop will address selected aspects of our evolving field from the perspective of younger specialists, hopefully leading to enlightening discussions. As in previous years, a session will be devoted to the oral presentation of the abstracts submitted for the travel awards by pre-doctoral and post-doctoral fellows.

For additional information, contact:

Leslie Lublink e-mail: lublinkl@ohsu.edu

Workshop on Inborn Errors of Tetrahydrobiopterin Metabolism

Diagnosis - Treatment - Follow-up - Outcome - Molecular genetics, Genoa, Italy, Tuesday, 7 September 1999, 09.30-12.00. For further information contact:

Dr. Nenad Blau,
University Children's Hospital,
Division of Clinical Chemistry and Biochemistry,
Steinwiesstrasse 75, 8032
Zürich / Switzerland,
Tel.: +411 266 7544, Fax: +411 266 7169, E-mail: blau@kispi.unizh.ch

The Human Genetics Society of Australasia annual scientific meeting

will be held at the Australian Technology Park in Sydney from 29th August to 1st September.

The Australasian Society for Inborn Errors of Metabolism meeting will run from 1st to 2nd September at the same venue with a joint plenary session with HGSA on the 1st. Programme details, registration information and call for abstracts will be posted on the HGSA web site: www.racp.edu.au/open/hgsa.htm

13th Annual symposium of the APS (Arbeitsgemeinschaft für Pädiatrische Stoffwechselstörungen)

to be held from 11-12th march 1999 in Fulda, Germany.

Topic: "Diagnosis of inborn errors of metabolism - Possibilities and limitations". Organisation and information:

*PD Dr. Ertan Mayatepek,
Division of Metabolic Diseases,
University Children's Hospital,
Im Neuenheimer Feld 150,
69120 Heidelberg,
Germany.
Phone: +49-6221-562311; Fax: +49-6221-563651;*

EUROMIT 4 - 4th European Meeting on Mitochondrial Pathology

will be held at Queens' College Cambridge between 16-19 September 1999.

The meeting is intended to cover all aspects of mitochondrial disease. Further details can be accessed through our website (<http://www.gen.cam.ac.uk/euromit>), e-mail: (r.n.lightowers@ncl.ac.uk) or by writing to:

*Euromit,
Dept of Neurology,
Medical School,
Framlington Place,
Newcastle upon Tyne
NE2 4HH,
UK.*

Workshop "Inherited Metabolic Diseases" ("Dedicne metabolicke poruchy")

on May 26-28 1999 in Litomyšl (East Bohemia), Czech Republic. Main topics: Homocystein,

Neurodegenerative IMD, Hepatopathy in IMD, Varia; Lectures and Posters Sessions; Language: Czech, Slovak, English.

Contact Erika Marklov, MD: marklova@lfhk.cuni.cz

5th International symposium on Mucopolysaccharide and Related Diseases.

Vienna, March 18-21, 1999. Further details:

*Tel: ++49 (0)621 41 06 152, Fax: ++49 (0)621 41 06 202,
email: mps99@meet-ideas.de*

Parent and support groups

UK Association for Glycogen Storage Disease's 1999 annual Family Conference

Saturday, 2nd October 1999, Manchester, The Copthorne Hotel, Clippers Quay, Salford Quays, Manchester M5 2XP. Further information from:

Mrs Ann Phillips abphillips@ukagsd@u-net.com

Research Trust For Metabolic Diseases In Children.

Annual Conference. Stakis Hotel Bromsgrove UK, 16th -17th October 1998. For further information

tel: (0)1270 250221. Fax: (0)1270 250244.

The Society for Mucopolysaccharide Diseases.

Annual Conference. Stakis Country Court Hotel, Northampton, UK, 10th-12th September 1999. Further information from:

*The Society for Mucopolysaccharide Diseases,
55 Hill Avenue,
Amersham,
Buckinghamshire,
HP6 5BX.
Tel: (0)1494 434156. Fax: (0)1494 432252. email: mps@btconnect.com*

NSPKU 26th Annual Conference

5th-7th March 1999, Swallow Hotel, Gateshead. Further details from:

*NSPKU conference,
Swallow Hotel,
High West St.*

Gateshead,
Tyne and Wear
NE8 IPE.
Tel (0)191 477 1105

European Mitochondrial Disease Network (EMDN)

For further information contact:

+44 (0)1744 851235 (fax and tel), email: pprestonmiyonet@netcentral.co.uk

Japanese Society for Inherited Metabolic Disorders

41st Annual Meeting, Tokyo, November 12-13, 1998

In November last year I had the very enjoyable experience and honour to deliver a special lecture at the annual symposium of our sister society in Japan. The meeting took place in a very traditional building serving both as a conference centre and also catering for wedding ceremonies. The scientific programme comprised mainly 2 parallel sessions of 125 free communications with one short symposium on Hox genes and two plenary lectures, one on homeotic proteins, molecular architects of body patterning by William McGinnis from the University of California, San Diego and my presentation on Homocysteine disorders. Apart from the last two of these, all communications were in Japanese, so that my understanding was restricted to the titles in the abstract book in English and also the English expressions which are liberally used throughout the abstracts. The content of presentations paralleled closely one of our own symposia ranging from molecular genetic studies to clinical data and covering the whole spectrum of inherited metabolic diseases.

As well as a traditional Japanese meal, with approximately 20 council members, I also took part in the general business meeting with the selected representatives (about 50) of the membership which numbers more than 600. This gave me the opportunity to introduce the plans for the 2000 meeting in Cambridge which were enthusiastically received as expressed by the promise of support and large attendance. A personal highlight was to be elected as an honorary member of the Japanese SIMD.

In addition to the meeting itself my wife and I were able to take the opportunity to extend our visit, taking in many of the attractions of Tokyo itself as well as trips further afield to Kamakura, Nikko, Hakone, Kyoto and Nara. We enjoyed the special Japanese mix of ancient culture, with the many, many temples, and modern developments as exemplified by the ever punctual Shinkansen bullet train system.

I wish to acknowledge the very warm hospitality which we enjoyed from all our hosts, but especially Prof. Eto, Prof. Matsuo, Dr. Ida, Prof. Kitigawa and Prof. Tada. The fruitful discussions on many matters of common interest, especially in the organisation of the joint International Congress confirmed the value of my visit in furthering the future co-operation of our two societies.

Brian Fowler

ERNDIM-QUESTIONNAIRE for European Test Directory

Under the umbrella of the ERNDIM Biomed2 project we wish to update the information on laboratories involved in the diagnosis and follow up of inborn errors of metabolism. We are fully aware that a directory (European Directory of Diagnostic Laboratories, P.Durand & E.Levi, Eds) has already been established in 1991 and that our British colleagues supply such UK- information on the WEB.

In order to update the directory and expand it to the European continent a questionnaire has been prepared based on the structure and abbreviations used in the U.K. Directory of Laboratories for Inborn Errors of Metabolism which can be found on the SSIEM Homepage <http://www.ssiem.org.uk> This questionnaire has been circulated to participants of the ERNDIM quality control schemes. Other laboratories which wish to be included but have not been sent a copy should contact either myself (Email, fowler@ub.acu.unibas.ch) or Claude Bachmann LCC CHUV, CH- 1011 Lausanne, Switzerland. (Email, cbachman@hola.hospvd.ch)

The questionnaire concerns information on the laboratory itself, experience accumulated by the laboratory when performing the tests listed in various tables, the amount of workload, the methods used, and information on chemicals (standards) not commercially available and deals with the tests actually performed in your laboratory.

If you choose to send the relevant pages of the questionnaire by email, please use Word 6 or 7 for PC or the rtf format for the documents attached (MIME protocol).

The deadline for ERNDIM laboratories has already expired so please act as soon as possible.

Brian Fowler

Dietitians meeting, SSIEM Genoa

This years dietetic meeting will be on dietetic issues in the adult inherited metabolic disease. Topics will include glycogen storage disease, MSUD, urea cycle disorders, homocystinuria and hyperphenylalaninaemia. For further details contact:

*Anita MacDonald,
Head of Dietetic Services,
Birmingham Children's Hospital,
Steelhouse Lane,*

Birmingham,

UK,

B4 6NH.

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