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# SSIEM Newsletter

Number 27 February 1998

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## Secretary's column

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1998 has arrived and with it severe gales, at least in Northwest Europe. Perhaps this is an omen of stormy times to come. However the SSIEM remains a bastion of stability as we move towards the end of this millennium. Our membership continues to grow in a quiet non-threatening way, with no danger of boom and bust cycles; our finances remain healthy despite the turbulent times in the financial markets; and our annual symposia continue to be a model of good organisation and good science. We look forward to the coming years in an optimistic mood and hope for yet better things to come. Most of us have survived the internal market, now we have to weather bird flu, global warming, BSE, and probably a few other natural and man made disasters.

This year's annual symposium is York, a medieval city encircled by well-preserved walls (largely 14th century), and famous for its magnificent Gothic cathedral, popularly known as York Minster. The meeting is being held at the University, and this will provide us with excellent lecture facilities. A more detailed report is included elsewhere in the newsletter.

I did hope to have a lively correspondence section in the newsletter. However despite my pleadings I have had almost no letters sent for publication. I contemplated writing some controversial diatribes myself, using a pseudo-name, but thought that would probably not be considered ethical. In any case Metab-L provides a much more immediate way of communicating. However I would still be very pleased to receive letters from members.

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## Report from Council

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Council continues to meet regularly during the year. The planning of the symposia remains the major work of council but other important items have included reports from the treasurer and editor of the JIMD. The Council's responsibility is to the members of the SSIEM. It is therefore imperative that Council is aware of the opinions of members. Using email it is

Wolf Endres and Garry Brown retired from Council in September. They have provided great service to the SSIEM and Council thanks them for their efforts. Garry Brown will be involved with the organisation of the 2000 symposium in Cambridge. Dr Jane Collins, Paediatrician at Great Ormond St Hospital and

now exceptionally easy to contact either corresponding or council members. Please do!

Professor Kurt Ullrich from Hamburg were elected to Council at the last symposium.

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## Corresponding members

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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, Canada  
Prof Y. Eto, Japan  
Prof M Ugarte, Spain  
Dr S Schweitzer, Germany  
Dr B Wilcken, Australia

We welcome Susan Schweitzer who replaces Kurt Ullrich as corresponding member for Germany.

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:

The Willink Biochemical Genetics Unit,  
Royal Manchester Children's Hospital  
Manchester  
M27 4HA  
Fax: + 44 161 727 2137  
E-mail: [John@jhwalter.demon.co.uk](mailto:John@jhwalter.demon.co.uk)

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*Dr J H Walter*

*Honorary Secretary*

*January 1998*

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## 35th Annual Symposium

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Despite being in the same year as the International Symposium in Milan the 35th Annual Symposium in Gothenburg still attracted over 340 active delegates. The organisation and hospitality provided by the local organisers and lead by Elizabeth Holme were excellent.



The picture represents the high point of the meeting with our chairman leading a karaoke session and rendition of the SSIEM's boating song.

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## Future joint International meetings of the SSIEM with the SIMD, JSIMD and International Congress

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The question of joint meetings of our society together with the International Congress on Inborn Errors of Metabolism, which was first held in 1977, was often raised in the past and indeed in 1980 the SSIEM forsook its meeting and participated in the International Congress of that year held in Switzerland. For several reasons this was considered unsuitable by many of our members and since then we went our separate ways. The chapter was reopened in May 1994 at the International Congress in Milan when we were confronted with the question from many participants, including many SSIEM members, why we could not combine our meeting with the International Congress. This was prompted by the pressure of having to choose between two major international meetings in one year in the light of increasing restrictions on finances and time.

Soon afterwards discussions began between ourselves and the Presidents of the SIMD and Japanese SSIEM and the committee of the International Congress resulting in the proposal for a joint meeting in the UK in the year 2000 which was accepted at the AGM of 1995 in Toledo. This meeting was originally envisaged in Oxford but due to the lack of a suitably large lecture theatre has now been changed to Cambridge. This agreement led to further discussions to formalise the future organisation of the joint International Congress to guarantee high quality of scientific content, venue and organisation. A plan was formulated by representatives of the three societies and the IC committee in May of last year at Vienna. Following intense discussion in Council and with Corresponding Members the plan was accepted unanimously at the AGM in Gothenburg on my recommendation. (See minutes of the AGM for more details).

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Briefly the plan is to hold a joint international Congress every three years organised in rotation by either the SSIEM or SIMD or Japanese or Australian societies. Every other one of these of these will be organised by the SSIEM in Europe, i.e. in 2000, 2006, 2012 etc. and in 2003, 2009, 2015 etc. the international meeting will be organised by one of the other groups. A praiseworthy aim is that the sponsoring society will endeavour to provide a significant number of travel grants and to minimise registration and accommodation costs to maximise access to young researchers in the field and of course a clash of meetings in the same year will be avoided. The responsibility for planning and allocating the sites and organisers for the future international meeting will be taken by a Joint Executive Committee.

I hope you will all agree with me that the advantages of closer contact and more opportunity for collaboration with our colleagues world-wide and the avoidance of a clash of meetings far outweigh any disadvantages which might be apparent. We now face the considerable challenge of trying to set new standards for the future International Congress and I am convinced that with the support of all interested parties this will be guaranteed.

I look forward to welcoming as many members of the various societies as possible, to Cambridge in the year 2000, a meeting which will set the stage for further joint meetings in the new millennium.

Finally I wish to take this opportunity to thank all the many members, colleagues in Council and Corresponding Members for their valuable contributions to the debate on this important topic.

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## JIMD moves to more frequent publication

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Every author wants his/her paper published the moment it is finished. The average time taken for publication in *Journal of Inherited Metabolic Disease* is no longer than in many other specialised journals, and a good deal shorter than in some, but our quarterly publication schedule inevitably has delayed some papers.

This year we will be increasing our publication frequency to give six regular issues containing full papers, case reports, short reports, etc. as opposed to four previously.

There will be two further issues devoted to the proceedings of the SSIEM Annual Meeting: one to Short Communications and the other to Symposium Reviews. The SSIEM Meeting abstracts will continue to be produced as a supplement and other supplements dealing with special topics will appear from time to time. More frequent publication will give us added flexibility when assembling issues for press and reduce the average time taken for an accepted paper to appear in print.

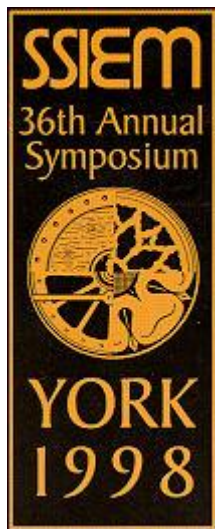
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***Dr Rodney Pollitt***

***Editor-in Chief***

***JIMD***

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## 36th Annual Symposium, 1st-4th September 1998

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The SSIEM this year will take place in the historic University city of York. The program has a broadly neurological theme with contributions on neurotransmitter disorders, glutaric aciduria type 1, Canavan disease and Batten disease. In addition, there is a morning devoted to fat oxidation defects and carnitine metabolism. The Tuesday afternoon workshop is to be a debate around the topical issue of "mass screening for metabolic disease". This year, as something of an experiment, we are hoping to include more short presentations from members by trying "chaired poster talks" where up to 40 posters will each be summarised using 2/3 overhead transparencies in carefully chaired sessions. We hope that there will be something to interest everyone.

The Social program will be no less interesting with the Conference dinner set in the well known National Railway museum and opportunity on Thursday afternoon to explore the countryside around York or visit this interesting medieval town.

Please register early and come ready to contribute. Further information, including the Final announcement is available on the SSIEM web site.

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***Dr M Henderson, Leeds &  
Dr J Bonham, Sheffield***

***On behalf of the Organising Committee***

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# ERNDIM

## (European Research Network for evaluation and improvement of screening, diagnosis and treatment of Inherited Disorders of Metabolism)

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Most members of the SSIEM already know that ERNDIM acts as a commissioning organisation for quality assurance schemes related to the laboratory detection of inherited metabolic disorders. The schemes available include : quantitative organic acid analysis, quantitative aminoacid analysis, qualitative organic acid analysis and a "special assays" scheme which includes carnitine, intermediary metabolites, very long chain fatty acids and other compounds. &NBSP A proficiency scheme is also available in the Benelux countries and similar schemes are planned for the rest of Europe. One hundred and seventy-six centres from 25 countries take part in these schemes. The existing activities are now run without any EU subsidy and it is hoped that the impetus gained from the support received as part of the successful BioMed 1 grant will continue to benefit QA in this area.

This year we were pleased to hear that an additional grant under the BioMed 2 project had been awarded. The plans for this work include: continued educational activities, work on developing calibration standards, a study to compare reference values for different metabolites, the wider development of proficiency testing schemes, the production of a regular Newsletter and organisation of meetings to discuss the efficacy of QA in this area, the comparison of analytical methodologies and an update of the European Directory. &NBSP This is an ambitious 2 year program with individual team leaders commissioned to deliver different aspects of the project. Those involved are all SSIEM members and ERNDIM undoubtedly benefits from close links with this society. If you have any views or suggestions - or offers of help please contact either myself or Baudouin Francois (Dr L Willems Institute, Universitaire Campus, B3590 Diepenbeek, Belgium).

Further information about ERNDIM is available on the SSIEM web site.

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*Dr J Bonham*

*Chairman*

*ERNDIM*

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## News from Australia

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Australia seems set to join the headlong rush into tandem mass spectrometry. Here in **Sydney** we have an instrument at the New Children's Hospital which we are using to screen 95,000 babies yearly, as well as having time to pursue a number of developmental projects. We understand that two other states may follow suit in the coming year. In Melbourne the drought of metabolic physicians in the Victorian Clinical Genetics Service has been broken, with the arrival last year

The Adelaide Women's and Children's Hospital Chemical Pathology department is seeking to appoint a chief hospital scientist with expertise in peroxisomal disorders, and this position was recently advertised in Nature. Professor Alf Poulos from the same department has retired to pursue a career in legal consultancy in relation to biomedical research. And if you want an excuse to come to Australia, the Annual Scientific Meeting of the little sister organisation, the ASIEM (**Australasian**

of Dr Avihu Boneh, from Hadassah University Hospital, Jerusalem. This year Dr Steve Kahler, from Duke University, will take over as Clinical Director of the Genetics Service, and this will further strengthen metabolic medicine in Melbourne.

**Society for Inborn Errors of Metabolism**) is in Melbourne this year in the first week in July, in conjunction with the meeting of the Australasian Human Genetics Society meeting. More information from Dr David Thorburn (address in the handbook).

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**Bridget Wilcken**

**Sydney**

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## Meetings diary

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### Future SSIEM symposia 1998 to 2002

1998 York (1st-4th Sept)  
1999 Genoa (7th-10th Sept)  
2000 Cambridge (13th-17th Sept)  
2001 Prague  
2002 Dublin

### 10th International Conference on Pteridines and Related Biogenic Amines

Snowbird, Utah, USA

March 8-13, 1998

For further information:

Dr. Robert A. Levine  
William T. Gossett Neurology Laboratories  
Henry Ford Health Science Center  
1 Ford Place, 4D Research  
48202 DETROIT, IL, USA  
Fax: 001 313 874 4570  
E-mail: rlevine1@smtpgw.is.hfh.edu, or URL <http://www.unizh.ch/~blau/conf.html>

**The Gaucher Vereniging Nederland** (the Dutch Gaucher Society) will celebrate its 15th anniversary by holding a symposium on April 24, 1998 in the Ernst Sillemhoeve, Lage Vuursche, The Netherlands. The symposium is entitled 'Gaucher disease - opening up new horizons'. During the meeting an overview will be given of clinical and biochemical aspects of Gaucher disease. Furthermore, results obtained with enzyme therapy in The Netherlands will be presented and discussed. The language will be Dutch. An abstract book with English summaries will be available. The contact person is Mrs. M. Guijt, chairperson of the symposium committee, Ruitercamp 155, 3992 BZ Houten, The Netherlands, Tel/Fax +31 30 6374417.

**The 4th Fatty Acid Oxidation and Ketogenesis Conference** will be held from Friday, 3rd April to Monday, 6th April 1998 inclusive, at the Institute of Child Health & Great Ormond Street Hospital for Children NHS Trust, in central London. One of the sessions will be for Inborn Errors of beta-oxidation and ketogenesis.

There is a conference web page at: <http://www.ich.ucl.ac.uk/units/faoxk.htm>. Interested parties should e-mail [s.eaton@ich.ucl.ac.uk](mailto:s.eaton@ich.ucl.ac.uk) (or FAX +44 171 404 6181) as soon as possible.

**Homocysteine Metabolism. 2nd International Conference.** April 26-29, 1998. University Auditorium and Conference Centre, University of Nijmegen, The Netherlands. For further information contact the congress secretariat: Conference Agency, University of Nijmegen, M. Majoor-Tempelmann, PO Box 9111, 6500 HN Nijmegen, Tel: +31 24 361 22 32. Fax: +31 24 356 79 56. Email: [M.Majoor@buro.kun.nl](mailto:M.Majoor@buro.kun.nl)

**Association for Glycogen Storage Disease (United Kingdom). Annual Conference.** Saturday 3rd October 1998. Moat House Hotel, Oxford. Further details from: Mrs Anne Phillips, 9 Lindop Road, Hale, Altrincham, Cheshire WA15 9DZ. Tel: 0161 980 7303 email: [ae314@dial.pipex.com](mailto:ae314@dial.pipex.com).

**4th Meeting of the International Society of Neonatal Screening,** 13th -16th June 1998, Stockholm, Sweden. For further information contact Agne Larson, PKU Laboratory, Huddinge University Hospital 141 86 Huddinge Sweden. Tel: +46 8 585 813 90, Fax: +46 8 585 813 95.

**7th International Congress on Neuronal Ceroid-Lipofuscinoses,** 13<sup>th</sup>-16<sup>th</sup> June 1998, The University of Texas Southwestern Medical Centre, Dallas, Texas, USA. Further information from Tel: +214 648 2166, Fax: 214 648 2317. web site: [http://www.swmed.edu/home\\_pages/cme/cermain.html](http://www.swmed.edu/home_pages/cme/cermain.html)

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## Workshop on inborn errors of the $\gamma$ -glutamyl cycle

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At the 36th Annual Symposium of the SSIEM in York a workshop on inborn errors of the  $\gamma$ -glutamyl cycle (glutathione metabolism) will be held on **Tuesday, September 1, 1998 at 10-12 am.**

This will be an opportunity for SSIEM members and others to update their knowledge on clinical signs and symptoms, diagnosis and management of patients with defects in  $\gamma$ -glutamyl cysteine synthetase, glutathione synthetase,  $\gamma$ -glutamyl transpeptidase and 5-oxoprolinase or with other forms of 5-oxoprolinuria.

Admission is free. We are interested in getting in contact with as many investigators as possible who have patients with disorders of the  $\gamma$ -glutamyl cycle. If you have patients with defects in the  $\gamma$ -glutamyl cycle please let us know. **Please register no later than May 15, 1998.**

### Preliminary program

- Introduction (Prof. Agne Larsson)
- Enzymology of the  $\gamma$ -glutamyl cycle (Dr. Mary E Anderson)
- Clinical picture (Dr. Ellinor Ristoff)
- Molecular genetics (Toxicologist Runa Njålsson)
- Antioxidants (Prof. Elisabeth Trimble)
- Treatment (Dr. Mary E Anderson)
- Case presentation and discussion
- Conclusion/ Summary (Prof. Agne Larsson)

For further information please contact. Prof. Agne Larsson

Dept. of Pediatrics

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S-141 86 Huddinge  
Sweden, Tel:+46 8 585 814 97, Fax:+46 8 585 874 40  
e-mail: gne.larsson@klinvet.ki.se

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## Letters

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### European concerted action "peroxisomal leukodystrophy"

The European CA "Screening, Prevention, Treatment and Pathogenesis of Congenital Peroxisomal Diseases" is in its second year now. The consortium includes 23 clinics or laboratories in 10 countries that are the partners financed by the European Union. Other centres in Europe, Israel and the US are collaborating with the consortium. The CA has held plenary meetings in Athens, Cardiff (1996), Vienna and Göteborg (1997).

One of the aims of the CA is the registration of all peroxisomal patients in Europe; a one page registry form, including diagnostic categories and a code number, was developed and is also available on retrievable computer file (IBM compatible and Apple Macintosh). New peroxisomal patients can be notified to the national registrars

### **National Registrars**

#### **Austria**

Dr. B. Molzer, Vienna  
Tel 43 1 40400 5501  
Fax 43 1 40400 5511  
E-mail : Hans.Bernheimer@akh-wien.ac.at

#### **Belgium**

Prof. Dr. F. Roels, Gent  
Tel 32 9 224 02 24 (301, 308)  
Fax 32 9 225 94 52  
E-mail : frank.roels@rug.ac.be  
S. De Bie Fax 32 9 240 4970 - Tel 32 9 240 2417 -  
E-mail : sylvia.debie@rug.ac.be

#### **France**

Prof. P. Aubourg, Paris  
Tel 33 1 4048 8249  
Fax 33 1 4048 8340  
E-mail : paubourg@infobiogen.fr  
Prof. J.-M. Saudubray, Paris  
Tel 33 1 44 49 48 52  
Fax 33 1 44 49 48 50

## **Germany**

Dr. J. Gärtner, Duesseldorf  
Tel 49 211 811 6184  
Fax 49 211 811 8757  
E-mail :gaertnj@uni-duesseldorf.de

## **Greece**

Dr. H. Michelakakis, Athens  
Tel 301 77 09 241  
Fax 301 77 00 111

## **Italy**

Mr. Ubaldo Caruso, Genova  
Tel 39 10 5636582  
Fax 39 10 3773210  
Dr. G. Uziel, Milano  
Tel 39 2 239 4447  
Fax 39 2 266 4236

## **The Netherlands**

Dr. Peter Vreken, Amsterdam  
Tel 31 20 566 2268  
Fax 31 20 691 7735

## **Portugal**

Dr. M.-C. SA Miranda, Porto  
Tel 351 2 609 2404 or 600 4940  
Fax 351 2 609 2404  
E-mail : samir-ue@individual.Eunet.pt

## **Spain**

Dr. M. Giros, Barcelona  
Tel 34 3 227 56 00 Ext 7584 Fax 34 3 227 56 68  
E-mail : mgiros@medicina.ub.es

## **United Kingdom**

Dr. G. Besley, Manchester  
Tel 44 161 727 2143  
Fax 44 161 727 2137

## **See SSIEM members handbook for full addresses**

As there are no official registrars in the other European countries, contact the colleagues of your choice, or the general co-ordinator.

Other tasks of the consortium are: follow-up study of X-ALD either untreated or treated with GTE/GTO oil (in coll. with ALD-International Research Group, Berlin and Baltimore); clinical trials of docosahexaenoic acid treatment of DHA-deficient patients; complementation of fibroblasts, and mutation analysis.

Forthcoming national meetings are in Barcelona (Febr. 26th) and Athens (May 8-9th). Next plenary meeting will be in York (SSIEM satellite meeting). A list of internal documents is available from the coordinator.

## Stable isotope standards

Dear colleagues,

Frequently I receive requests to supply various unlabelled and stable isotope labelled compounds for use as standards in GC/GCMS methods or as substrates in enzyme studies. In the past I have made available a number of peroxisomal standards (deuterated VLCFA, phytanic and pristanic acids) and a series of deuterated and unlabelled acylcarnitines.

Now I have the intention to respond to the wants for acylglycines. The synthesis of a series of unlabelled, saturated acylglycines is in full production now, ranging from C<sub>2</sub> to C<sub>16</sub> including pivaloyl-, isobutyryl- and isovalerylglycine. Suggestions for others are welcome. For stable isotope labelled acylglycines there are several options.

a)

Labelling with deuterium in the acyl part. This affords acylglycines with at least triple labelling which is ideal for internal standards in GCMS. However, this would also imply the purchase of the whole series of individually deuterated acids which in many cases is very expensive.

b)

Triple labelling in the glycine part, using very expensive [<sup>13</sup>C<sub>2</sub>,<sup>15</sup>N]glycine and coupling with - relatively inexpensive - unlabelled acyl groups.

c)

Synthesis of [<sup>13</sup>C<sub>2</sub>]acylglycines by coupling [<sup>13</sup>C<sub>2</sub>]glycine and unlabelled acyl moieties which is a compromise between costs and optimal labelling.

I have not yet made my decision which option to choose for the labelled acylglycines.

The eventual prices for the compounds form an important aspect, however the choice for a labelling pattern is also determined by the specific application and the analytical techniques applied. You are invited to let me know if you are interested in obtaining acylglycines, including an indication of the amounts wanted, and to give your suggestions for the labelled ones.

To reduce the costs for synthesis by minimizing overproduction, which is of special importance if you require a large number of compounds, I want to collect the requests for these compounds in advance. Next I can give an indication of prices to the respondents, after which definite orders can be placed. Those of you who have participated in the fatty acid project and/or the acylcarnitine project will recognize this approach. Please specify your wishes via letter, fax or e-mail.

[Additional information](#) about several other standards is available at the internet:

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Amsterdam The Netherlands tel: 31 20 4442883 fax: 31 20 4440305 e-mail: [hj.tenbrink@azvu.nl](mailto:hj.tenbrink@azvu.nl)

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