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# SSIEM Newsletter

Number 30 August 1999

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## Reflections on changes seen as Chairman

by *Brian Fowler*

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My six years as Chairman have seen the activities of the SSIEM flourish, profiting from the sound financial basis and the new constitutional changes established thanks to the efforts of our previous Chairmen and their colleagues in Council.

This has seen a distinct change in the flavour of Council meetings with less need to devote time to administrative matters towards more discussion of the scientific content of future symposia and recently we have even become slightly political with the production of recommended syllabuses for training in inherited metabolic disease.

### Changing membership

One important development during my period in office has been widening of the representation of Council and Corresponding Members to reflect the increasing non-British membership. Steps taken include the almost doubling of the number of Corresponding Members with representation now of all continents. Importantly there have been increasing efforts to involve Corresponding Members in the running of the Society by, for example, distribution of minutes of all council meetings to them and more canvassing of their views on specific issues. Furthermore Council itself has been strengthened by including two members from mainland Europe, presently from the Netherlands and Germany

I must say as a British person who translocated to a middle European country I can vouch for the advantages of the wider view this has brought to addressing the issues which confront Council and personally I have appreciated the new accents heard in the council meetings themselves. On the other hand the benefits of the concentration of the administration of the Society in the U.K., in particular the close link to the running of the Journal of Inherited Metabolic Disease, remain strikingly evident in spite of the changing nature of the character of membership. When I took over as Chairman in 1993 the total membership was less than 800 with the large majority from the U.K. Today the number of British members remains similar but the total membership has increased significantly to almost 900 and the Treasurer confidently informs me that he expects a total membership of 1000 by next year.

### The size of the annual symposium

The increase in the size of the annual meeting has been the most obvious change in the last six years and is one which will continue to challenge our successors in Council. In Manchester in 1993 the size of the conference

I am sure I speak for many in stating that we should try to keep the format of no parallel sessions, a limited number of posters exhibited for the whole of the meeting, evening events for everyone together and an atmosphere which

centre forced us to limit the participant number to 300. Subsequently participation has increased dramatically so that in York last year we were severely tested by the attendance of more than 600 delegates. This high number of participants is clearly to be seen as confirmation of our success and a matter of pride but it also produces a number of issues to be solved in the future. As well as the organisational difficulties of finding large enough facilities for lectures, cheap accommodation and social events, how can the special nature of the SSIEM meeting be maintained?

colleagues meet informally and discuss and plan the collaborative studies so vital in our field. . We have already tried parallel workshops on the first afternoon, poster walks, poster talks and the publication of the abstracts in advance as an issue of the JIMD in addressing the problems of larger meetings. Finally in relation to our annual symposium we can take pride in the increase of the travel and registration grants for younger colleagues with lack of financial support which increased from a total of £3000 in 1993 to £6800 for this year in Genoa.

## Future collaboration with other Societies

Discussions began at the International Congress in Milan in 1994 to try to bring together the main Societies to establish of a firm basis for the future organisation of the Int. Congress. A major outcome of my period in office, following detailed and sometimes frank discussions with the Officers of the SIMD, ASIEM and JSIMD and the Chairman of the International Congress, is the plan that future International Congresses will be organised by one of the Societies beginning with the Cambridge 2000 meeting. The guidelines for this future co-operation have been noted in previous Newsletters and AGMs and at this point I merely wish to say that we are looking forward to next year's Symposium with great optimism and hope to set the standard for future International Congresses held on this new basis. We also look forward to a strengthening of bridges with our sister societies which will clearly be of benefit to of all members of our societies.

*Brian Fowler*

In closing, a reflection of the scientific content of our meetings and publications confirms the enormous advances made in our field during recent years and these owe a great deal to the co-operation between workers of different disciplines from different countries. I believe our Society's activities, in particular the annual symposium play an important role in nurturing such co-operation and long may this continue.

I wish to thank most sincerely all members and colleagues who have help to make my period in office, first as Secretary then as Chairman so enjoyable. It has been a great honour to fulfil this role and I step down with much confidence for the future and I wish my successor an equally rewarding and enjoyable experience in what is likely to be an exciting period in the further development of the SSIEM.

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## Publishing in the JIMD - *a practical guide.*

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The Journal of Inherited Metabolic Disease is a major element of the SSIEM's contribution to fostering the study of

The Society has always had a strong commitment to improving the care of patients with metabolic disease: indeed the first meeting was organised partly in the

inherited metabolic disorders. The association is close: SSIEM members receive the Journal free as part of their subscription and three issues a year (Abstracts Supplement, and Short Communications and Reviews issues) are devoted to the Society's annual symposium. However, the aims of the Society, as well as the economics of publishing, require JIMD to be a fully-fledged publication in its own right rather than just a house journal. The majority of its output comes as submitted papers from scientists throughout the world, whether SSIEM members or not, and the publisher, Kluwer Academic Press, maintains substantial sales to institutional subscribers

hope of persuading the authorities in Sheffield to introduce newborn screening for phenylketonuria (K. S. Holt, personal communication, September 1987). Additionally, the Journal's founding editor, Noel Raine, believed that we had a moral duty to encourage research and service provision in the less-developed parts of the world as well as at home, an approach reflected in the geographical spread of the Editorial Advisory Board that he assembled. However, our subject also has a strong academic base and the current editors have the task of balancing the evangelical approach against the need to remain competitive in the "market", attractive to potential contributors increasingly harassed by bibliometric research assessment exercises\* as well as to our more general readers

What are the editors looking for in a submitted paper (or even a poster abstract)? Novelty, interest and impact is the simple reply, but what does this mean in concrete terms? In practice it is much easier to describe what we **don't** want.

**JIMD** is an international journal so that a paper must have more than just local interest: is the message potentially generalisable?

**P**redictability and triviality are the two biggest turn-offs. "The first case of aspartylglycosaminuria from the Outer Hebrides" has both and goes straight into the bin: sporadic cases of every genetic disease that has ever been discovered will turn up sooner or later in any but the most genetically isolated of populations.

'Amazing coincidence' papers such as "A case of argininosuccinic aciduria with a phenylketonuric twin" are also likely to receive short shrift.

**P**erhaps less obviously trivial is "First prenatal diagnosis of xyz-disease by mutation analysis using chorionic villus" but unless some special problem has been encountered does such a straightforward application of known technology really merit recounting

**T**he task of selection is made all the more difficult by increasing clinical activity in many parts of the world and the explosive growth of very detailed knowledge, particularly at the DNA level. It is evident that not all such new information is suitable for publication in the traditional form of a scientific paper. New mutations are particularly difficult to adjudicate. In theory, every position in every gene is susceptible to mutation so that new examples are both predictable and trivial. However, mutation patterns and frequencies, expression studies and genotype-phenotype correlations are worth recording and suitable ways of accumulating such data need to be found. Centralised collection, either the old-fashioned way or electronically via the Internet, is a prerequisite to useful synthesis. The recent review by Scriver and Nowacki (JIMD 1999; 22: in press, or, in an updated version with hypertext links to other sites, on the SSIEM web site (<http://www.ssiem.org.uk>) gives an inspirational view of how such resources might be developed and used. Some see the growth of internet data-bases and electronic publishing as foretelling the end of traditional paper-based publishing but the two will remain complementary for many years; once you have them on your desk, well produced hard copy publications are so much easier and quicker to read. Judging from citation statistics, papers in our field have a long useful life so that presentation and durability are at least as important as immediacy.

We should encourage interactivity and the most appropriate use of both media. Perhaps paper journals will soon be coming with bar-coded hypertext links in the reference lists. In turn, internet availability of facilities such as Medline is a great help in locating the papers that you want. For those with long runs of JIMD on their shelves but who can never quite remember how long ago it was that they saw that article by...., Kluwer have a searchable index of the Journal, accessible by hypertext link from the SSIEM site.

Wonderful isn't it

*RJ Pollitt*

\* Seglen PO. Why the impact factor of journals should not be used for evaluating research. *Br Med J* 1997; **314**: 498-502

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## Secretary's column

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Dr Brian Fowler retires as chairman of the society this year. Brian has devoted considerable time and effort to further the society, both as chairman and prior to that office as secretary. In this newsletter he reflects on the many changes that have occurred over the past decade, a period that has been characterised by a considerable increase in the interest in metabolic disease. The society is now approaching 900 members and has become truly international. Brian's stewardship has been highly successful and the society owes him a debt of gratitude for this. Brian has agreed to lead the organisation of the Cambridge 2000 meeting so we will continue to draw on his experience and expertise for at least a further year!

Dr Rodney Pollitt, editor of our journal, the JIMD is also standing down as editor in chief, although not until the end of this calendar year. There is wide recognition that the journal has shown a considerable improvement in quality and although this is of course dependent upon submitted articles, the editor plays a pivotal role in nurturing such developments. In the newsletter he discusses the role of the JIMD and provides a practical guide as to how to get articles submitted for publication. Now I know why my article "The first case of PKU in the village of Little Wollop" was rejected!

The following council members are due to retire in September: Brian Fowler as Chairman, myself as Secretary, Albert van Gennip and Peter Clayton. Kim Bartlett has served 3 years as Council member but is willing to stand for re-election for a further 3 years. Nominations for council should be sent to me at the Willink Biochemical Genetics Unit, Royal Manchester Children's Hospital, Pendlebury Manchester UK, M27 4HA, to arrive no later than 1st September 1999.

A number of corresponding members have also served their terms of office. An updated list will be published in the next newsletter.

It is proposed that Dr Mick Henderson takes over as secretary so provided that this is ratified at the AGM you will need to send material for subsequent newsletters to him (Dept of Clinical Chemistry, St James University Hospital, Leeds, UK, fax +44 (0)113 206 5971, email mp.cpsjuh@pop3.hiway.co.uk.).

I look forward to seeing as many of you as possible in Genoa.

*Dr J H Walter, Honorary Secretary, July 1999*

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## Meetings diary

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### Future SSIEM symposia 1999 to 2002

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

### 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](mailto:baumgj@mail.health.state.mo.us)

### 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,  
032 Zürich / Switzerland,*

Tel.: +411 266 7544, Fax: +411 266 7169, E-mail: [blau@kispi.unizh.ch](mailto:blau@kispi.unizh.ch), URL: [www.unizh.ch/%7Eblau/nblau.html](http://www.unizh.ch/%7Eblau/nblau.html)

### 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/euomit>), e-mail:

([r.n.lightowers@ncl.ac.uk](mailto:r.n.lightowers@ncl.ac.uk)) or by writing to:

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

**The 6th Congress and 1st Advanced Course of the European Society of Magnetic Resonance in Neuropediatrics (E.S.M.R.N.)** will be held in Marseille, France, January 19 - 22, 2000.

Topics :

- | Prognostic Expectations in Pré and Périnatal Anoxo-Ischemic insults
- | New Developments of NMR in Neuropediatrics
- | Epilepsies
- | Malformations
- | Metabolic Diseases

For information, contact :

*Charles Raybaud, Neuroradiologie,  
Hôpital de La Timone,  
13385 Marseille Cedex 5,  
France.*

Tel : 33 (0)4 91 38 65 26 ; fax : 33 (0)4 91 38 68 33 ; e-mail : [charles.raybaud@wanadoo.fr](mailto:charles.raybaud@wanadoo.fr)

or the Administrative Secretariat:

*MCO Congrès, 27,  
rue du Four à Chaux,  
13007  
Marseille,  
France.*

Tel 33 (0)4 95 09 38 00 ; fax : 33 (0)4 95 09 38 01 ; e-mail : [mcocongres@aix.pacwan.net](mailto:mcocongres@aix.pacwan.net)

[http:// www.seminaire.com/mco](http://www.seminaire.com/mco)

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## Parent and support groups

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### **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](mailto: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](mailto:mps@btconnect.com)*

### **European Mitochondrial Disease Network (EMDN)**

Mr Paul Preston, Director of EMDN, has been invited by the Russian Minister of Health to present a talk to parents and professionals at the Moscow Institute of Paediatrics and Surgery in November. EMDN are aiming to raise money for the Moscow Institute to provide them with reference journals and medical text books.

For further information contact:

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



## **Letters**



### **Gastrointestinal manifestations associated with GSD Ib/c**

Researchers at Washington University School of Medicine in St. Louis are looking for collaborators for an international study designed to investigate the gastrointestinal inflammation associated with glycogen storage disease Ib. A significant number of GSD Ib patients develop debilitating intestinal inflammation, which in many instances results in the intestinal phenotype of Crohn's disease. This study is designed to test the hypothesis that neutrophil dysfunction (in the setting of specific environmental influences) is responsible for the inflammatory phenotype in GSD Ib as well as idiopathic Crohn's disease. Accordingly this study will examine the similarities between Crohn's disease and the intestinal manifestations of GSD Ib; 1) by standard histologic, endoscopic, and radiologic diagnostic criteria, 2) by comparing patient responses to specific medical and surgical forms of treatment, and 3) by comparing expression of serologic and/or genetic markers (e.g. ANCA, ASCA). Investigator participation would involve filling out a short data form, providing copies of pathology, endoscopy and radiological exams, and collection of a serum sample for analysis. Individuals interested in participating or having additional suggestions are asked to contact Dr. Brian Dieckgraefe in the Division of Gastroenterology at Washington University School of Medicine by email ([dieck@im.wustl.edu](mailto:dieck@im.wustl.edu)) or if unavailable, by phone or fax (314-362-8940, fax:314-362-8959).

## **Alkaptonuria in adult patients: a multinational study**

The clinical course and prognosis in adult life for individuals with Alkaptonuria is largely unknown. We are currently designing a multi-national comparative study in adult patients. The aims of the study are to assess whether the association between age and profession with the prevalence of joint abnormalities in this patient group can be confirmed, to assess the risks of developing musculo-skeletal abnormalities and cardiovascular disease for patients and to evaluate possible differences in clinical manifestations in different countries.

The design of the study is an analysis of the known adult patients with alkaptonuria in a centre or region. The inclusion criteria are unequivocal biochemical diagnosis of alkaptonuria (increased homogentisic acid in urine) and age > 20 years. A detailed protocol is available on request. The aim is to include a total number of 40 patients. We would like to invite you to participate in this study.

Additional information can be obtained from the address below:

*Dr Harold W. de Valk MD PhD,  
Dept Internal Medicine F02.124,  
University Medical Center Utrecht,  
Heidelberglaan 100, 3584 CX,  
Utrecht,  
The Netherlands,*

Tel: 31-30-2506312, Fax: 31-30-2518328, E-mail: [H.W.devalk@digd.azu.nl](mailto:H.W.devalk@digd.azu.nl)

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## **SSIEM awards**

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The SSIEM awards are made each year for the best short communications presented at the annual symposium and published in the JIMD. Two prizes are awarded each year, one for the best clinical paper and the other for the best biochemical report. The following awards were made following the York meeting last year:

### **Clinical prize**

Oral phenylalanine loading profiles in symptomatic and asymptomatic gene carriers with dopa-responsive dystonia due to dominantly inherited GTP cyclohydrolase deficiency

*K Hyland, TG Nygaard, Trugman JM, KJ Swoboda, LA Arnold and SP Sparagana J Inher Metab Dis 22(1999) 213-215*

### **Biochemical prize**

Missense mutations in the phenylalanine hydroxylase gene (PAH) can cause accelerated proteolytic turnover of PAH enzyme: A mechanism underlying phenylketonuria.

*PJ Waters, MA Parniak, BR Akerman, AO Jones and CR Scriver J Inher Metab Dis 22(1999) 208-212*

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## **Cambridge 2000**

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As most of you will be aware the annual symposium in 2000 is combined with the 8th International Congress on Inherited Metabolic Disease. This meeting, although primarily being organised by the SSIEM is supported by the Australasian (ASIEM), Japanese (JSIEM) and North American (SIMD) societies. It is therefore going to be a bigger and different event from our usual symposia. The program, which starts on the afternoon of Wednesday 17th September and continues until midday on Sunday 17th, will consist of plenary session each morning with parallel sessions in the afternoon. The overall theme of the meeting is "basic science to clinical disease". Cambridge has excellent facilities and we anticipate a popular and stimulating meeting.

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