
SSIEM Newsletter

Number 28 August 1998

Secretary's column

Welcome to the society's 28th newsletter. This first half of the year is a relatively quiet time for the SSIEM since the annual symposia are held in September. However there is a lot going on behind the scenes particularly with regards to the planning of forthcoming meetings. The meeting in York is going to be particularly popular and fortunately the facilities are more than adequate to accommodate us all. Please do try and attend the annual general meeting on September 2nd and make your views known.

Report from Council

Council has met on two occasions since the publication of the last newsletter; at Sheffield Children's Hospital on 20th February and at Great Ormond Street on 26th June. The agenda for these meetings are circulated to corresponding members prior to the date in order to allow them to let me have comments.

These can then be discussed by Council. The minutes of each meeting are also sent to all corresponding members. In this way we hope to get as much input from members as possible. I am also very happy to receive phone calls, letters, faxes and emails from individual members.

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, Canada

Prof Y. Eto, Japan

Prof M Ugarte, Spain

Dr S Schweitzer, Germany

Dr B Wilcken, Australia

Dr O Elpeleg, Israel

Prof R Giugliani, Brazil

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:

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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

January 1998

36th Annual Symposium- update

The number of delegates attending the SSIEM Annual Symposium has been increasing steadily over the years and the York meeting seems as if it is set to be the biggest UK meeting so far with more than 450 delegates registered. Unfortunately, as a result of this "success" we have had to disappoint some individuals who submitted abstracts for poster presentation.

The SSIEM Council has always taken the view that 150 posters is probably near the upper limit for poster display and, with more than 330 abstracts submitted, a substantial number could only be accepted this year in abstract form. We attempted to assess submissions on the basis of their International relevance and scientific impact. Several submissions, while interesting, were presented in a way which emphasised their local relevance e.g. "Three cases of X condition in Y country".

It may be helpful, in future meetings, to emphasise those findings which have a more general and transferable message. In the meantime we hope that York will be a success and that the members attending

enjoy the meeting.

Dr JR Bonham

The Horst-Bickel Prize

This prize of 50,000 DM was founded in recognition of the pioneering work of Horst Bickel to promote European efforts in improving treatment of patients with inherited metabolic disorders.

The SSIEM has been closely involved in the promotion and administration of this very worthwhile prize since its conception in 1995. As well as our involvement in the establishment of the original guidelines, two of the 7 judges have been nominated each year from Council.

In its first year the prize was awarded to Dr. Sylvia Stockler for her work on the treatment of guanidino acetate methyl transferase deficiency.

However in the last two years the judges were unable to recommend a winner. They all felt that the guidelines were relatively restrictive and thereby limited the scope of work considered suitable for submission. In addition a more consistent form of content and format for the applications was considered desirable.

With this in mind the judges proposed a modification of the guidelines to encourage more applications. The guidelines for the prize have now been modified by Brian Fowler and Mr. Brehl of SHS International, Heilbronn together with the other 6 judges for the prize of 1997.

These modified guidelines and call for applications for 1998 should have been received by eligible centres and individuals by now. The main changes are that as well as demanding a more structured application, the criteria for submissions are widened for example to include basic information on a disease or description of a new disease, recognising that a disorder needs to be discovered before treatment can ensue. Importantly however, the requirement that the prize itself must be used to support further work on treatment itself ensures that the basic philosophy behind the prize will be adhered to. This philosophy encompasses the ideals of Professor Bickel and Mr. H. Brehl in that the enormously expanding knowledge at the molecular and biochemical level should be extended to yield improved treatment and outcome in our patients with inherited metabolic diseases.

The opportunity now lies before all European members of the SSIEM to submit suitable work for this worthy

award and we hope for a large number of applications for the prize of 1998.

Further information and details of how to apply contact:

Secretariat
Horst-Bickel-Foundation
Postfach 30 61
D-74020 Heilbronn, Germany

Dr Brian Fowler

Italian Society for Inborn Errors of Metabolism

The General Assembly of the Italian Society for the Study of Inherited Metabolic Disease (SISMME), held in Spoleto, on November 13th 1997, during the XII Annual Congress of the Italian Federation for the Study of Inherited Disease, has decided to maintain its independence, rather than join the newly founded Italian Society of Human Genetics. The Italian Society for the Study of Inherited Metabolic Disease will, therefore, continue, representing the Italian Referral Society in the field of biochemical, molecular and clinical aspects of inborn errors of metabolism.

Professor Enrica Riva – Department of Pediatrics, S Paolo Hospital, Milan has been re-elected unanimously as Chairman of the Society.

Members of the Council for the year 1998 are: Dr. Alberto Burlina, Padua; Prof. Roberto Cerone, Genoa; Dr. Barbara Garavaglia, Milan; Dr. Gaetano Sabetta, Rome; Dr. Franco Taroni, Milan. Secretary of the Society is Dr. Giacomo Biasucci, Milan.

The next SISMME Congress will be held in Milan, November 26th-29th, 1998.

Meetings diary

Future SSIEM symposia 1999 to 2002

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

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.

4th Meeting of the International Society of Neonatal Screening

13th -16th June 1999, 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.*

RTMDC Conference and Annual General Meeting

17th October 1998, The Stakis Hotel, Birmingham Road, Bromsgrove, Worcester, B61 0JB. Programme includes: Back to Basics – Metabolic Disease from the Beginning (Dr. Mike Patton); Parents Voice (Jil and Carolyn Philistin-Rabaud); Approach to New Treatments (Dr. Fran Platt, Dr. John Walter); Fatty Acid Oxidation Defects (Dr. Andrew Morris); Special Children: Special Conflicts (Tom Leimdorfer); Special Needs: A Dad's Perspective (Dave Rendall); Spreading the Word – Making your message count (Miss Celia Hall, Medical Editor – Daily Telegraph).

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:-

*Dr. O. Labudova
Rheinische Friedrich-Wilhelms-Universitat
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*

EAWIEM 5th Annual Meeting, Warsaw, Poland

6th – 10th September 1998. Scientific programme will include:

Mitochondrial cytopathies, B-oxidation disorders of fatty acid metabolism, Approaches to early diagnosis of inborn errors of metabolism, Genotype-phenotype correlation. The conference fee will be approximately US\$ 500 for active participants and US\$ 400 for accompanying persons for double occupancy. For further information please contact:

Dariusz Rokicki
Organizing Committee
Department of Metabolic Diseases
The Children's Memorial Health Institute
Al. Dzieci Polskich 20
04-736 Warsaw, Poland
Tel: 4822 157490 Fax: 4822-157489
e-mail: pronicka@czd.waw.pl

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 millenium. For further information please contact:-

ISNS 1999,
C/o Stockholm Convention Bureau
Box 6911, S-102 39 Stockholm
Sweden
Tel: +46 8 736 15 00
Fax +46 8 34 84 41
lsns@stocon.se

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 Worskhop 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

Annual Meeting of the Society for Inherited Metabolic Disorders (SIMD), March 1998

by Dr Brian Fowler

This year's annual symposium of our American sister society, the SIMD was held from March 15 – 18 this year in the attractive pacific coastal setting of the Asilomar conference centre which will be familiar to many of you as a most spectacular setting for the 1990 International Congress. I was fortunate to be able to attend this meeting, combined with a short visit to the Department of Human Genetics at the Mount Sinai School of Medicine in New York. This gave me the valuable opportunity to meet a large number of our North American colleagues and to compare the SIMD meeting with our own annual symposia. Incidentally the SIMD has nearly 250 members with considerable overlap of membership with that of the SSIEM. More information on the SIMD is available on the internet, linked through the SSIEM's own home page.

The meeting, ably organised by the President of the SIMD for next year, Greg Grabowski assisted by Leslie Lublink the meetings secretary, was attended by approaching 200 participants.

Reflecting on the meeting my main impression is of a very much more politically minded and less international event than our own symposia. For example a whole half day session was devoted to the organisation of biochemical genetic services and accreditation of responsible centres, in the light of developments away from specialist providers of services for diagnosis and management of our patients towards the generalists of the so called health management organisations.

In the business meeting, a much more lengthy affair than our AGM, I learned that the SIMD even has a committee responsible for providing expert opinion on relevant health related matters passing through government. In addition, at the instigation of Harvey Levy, the SIMD agreed to produce a policy statement on Newborn Screening urging state health departments to expand screening to include a wider range of disorders including MCAD. It is clear that such issues are much more appropriate for a national society than for a multi-national organisation such as our own.

Notwithstanding these important considerations the major emphasis of the meeting was the scientific programme which included a one day session titled "developmental perspectives on inborn errors". The first part of this session covered inborn errors of dysmorphogenesis with expert presentations on holoprosencephaly and the Smith-Lemli-Opitz syndrome linking cholesterol and hedgehog proteins, pycnodysostosis caused by lysosomal cathepsin K deficiency, cleidocranial dysostosis as a disorder of cell fate determination and aspects of FGF gene

The final morning of the meeting comprised short presentations by the eight winners of travelling scholarships for young investigators mirroring our own efforts to support attendance of younger workers at such symposia.

The last evening of the meeting began with an early dinner followed by an hour long Presidential address given by the incumbent for this year Greta Seashore.

The session for the 43 posters followed until late at night and a good and enthusiastic attendance was guaranteed by its taking place alongside the provision of adequate

function. The second part included developmental outcomes of inborn errors reviewing outcome and covering cognitive defects, neuropsychological testing and animal models for behavioural disorders.

supplies of liquid refreshment. These arrangements are in stark contrast to our own formal dinners, daytime poster presentations and much shorter speeches by our President and Chairman.

All in all it was a most enjoyable and rewarding experience and if you have the opportunity to visit the United States I can wholeheartedly recommend attendance at next year's meeting. This will be held at Lake Lanier Islands Resort outside Atlanta, Georgia from March 12-15. Details can be obtained from:

*Dr. Tony Velazquez,
Insituto Nacional de Pediatria,
Apdo. Postal 101-48, Mexico, DF
04530,
Phone and FAX: Int + 525 606 3489.*

Brian Fowler, Chairman SSIEM.



Letters



RTMDC Annual Conference, Bromsgrove Stakis Hotel – 17th October 1998 : A Parents View

I've been asked by several professionals if I can outline this year's Conference programme, so I've attached the Conference Agenda (see under Meetings Diary). For parents the cost is £25 each, for professionals the cost for the day is £40 and includes dinner, teas and coffees etc. Anyone paying before 1st May 1998, receives a 5% discount. Please contact the RTMDC direct if your are interested in attending. Some pointers:

Re the actual programme: Dr. Fran Platt is based at the Oxford Glycobiology Unit and Dr. John Walter is at the Willink Biochemical Genetics Unit, Manchester. I heard a similar talk re "Basics" from Dr. Pennock at

It is quite "normal" for all these specialists to chat with parents/carers in every conceivable nook and cranny of the hotel while both "sides" can meet together – corridors and odd offices, reception areas – coffee breaks and lunch chats – it is normal for often desperate parents – and, dare I say it, local professionals – to take the opportunity of "picking brains" – I've never known any specialist refuse to listen and advise – often at cost to themselves.

With such a full and varied programme, and such excellent speakers, and needy parents and local professionals, running to time can occasionally be a problem. I have had the privilege of seeing doctors – sometimes several from different top hospitals all at the same time, continuing to help folk who are struggling, way past "normal" limits. Medics coming to these conference are tremendously dedicated and committed – they care, they have usually given their time totally

Southmead, and it was superb – for ANY professional dealing with ANY metabolic disease, I suspect you won't better this information. I also have heard both Dr. Walter and Dr. Platt – and the new treatment being discussed will include Enzyme Replacement Therapy and Bone Marrow Transplant – again, they are terrific – the information is the latest available in the world. Tom Leimdorfer's mediation skills, and his sheer expertise re the LEA and current legislation statementing process is first class. Lastly, the Fatty Acid Oxidation seminar covers lots of diseases – the rare and more common – see programme details – I won't attempt to spell them myself!

free apart from travelling expenses (if any claimed) – and they do whatever it takes to help children like mine stay stabilised and carers given as much information as is currently available.

I also need to say that, in view of a recent meeting attending, having been to some of these conferences, the RTMDC's organisation is second to none – how on earth with the vast numbers involved, from often so many countries, they do this job year after year, I don't know – but they are a first class, dedicated staff, who will ensure your day isn't wasted. At these low prices, any professional wishing to help a child with any metabolic disease would learn a phenomenal amount.

Hope this helps – and hope to see some of you there, if you feel it would help you professionally.

A parent

For more information please contact:

*RTMDC,
Golden Gates Lodge,
Weston Road,
Crewe,
Cheshire,
CW2 5XN.*

Tel: 01270 250221 Fax: 01270 250244 Registered Charity NO. 283541

Letter of Gratitude

The medical care of Slovak children with inborn errors of metabolism has suffered badly because of the split of Czecho-Slovakia in 1993. All of a sudden our Czech friends and colleagues were "abroad" and we found ourselves deprived of appropriate technical equipment, medical experience and professional contacts with experts in Western Europe. Our Czech friends kept on supporting us, but the cooperation was now hampered by the newly drawn border.

At the same time, Slovak pediatricians' awareness of these disorders has improved, too. Inborn errors of metabolism were the major topics of the 1st Slovak Congress on Pediatrics held in 1997.

We would hereby like to thank all the people who have been helping us. We wish to express our unlimited gratitude to Mr. Helmut Brehl, the Executive Director of SHS Heilbronn, who kindly sponsored our membership in SSIEM as

However, in recent years we have made considerable progress in the field of inborn errors of metabolism. With advanced technology at our disposal, the diagnosis, monitoring and treatment of our patients has become more sophisticated than ever before.

well as our participation in important international meetings. He provided us with valuable medical literature, made it possible for our dietitian to be trained and granted his support to the organising of the Czecho-Slovak Congress on inborn errors of metabolism in 1996 in our country.

V. Bzduch and D. Behulova

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