

White Blood Cell Cystine Group Meeting

The first meeting of the WBC cystine group took place in Paris on 17 November. It was attended by ten leaders in the field of cystinosis, with representatives from the fields of biochemistry and clinical medicine. The members present included Prof. Erich Harms (Muenster), Dr. Brian Fowler (Basel), Dr. William vant'Hoff (Great Ormond St), Dr. Mick Henderson (Leeds), Dr. Neil Dalton (London), Prof. Michel Broyer (Paris), Dr. Bernadette Chadeaux (Paris), Dr. Christine Vianey-Saban (Lyon), Prof. Giorgio Federici (Rome) and Dr. Elena Levtchenko (Nijmegen).

The aim of the meeting was to unite all those involved in the treatment and care of cystinotic patients in the major European cities, in an attempt to achieve a standard method of WBC cystine measurement in all principal laboratories. Various issues were discussed, including differing opinions as to the methods of measurement, storage and transport of samples.

Prof. Erich Harms began with an update on the clinical and treatment aspects of cystinosis. He was followed by Dr. Brian Fowler, who commenced with an overview of the ERNDIM survey, and proceeded to discuss varying methods of cystine measurement. Dr.'s Dalton, Federici, Vianey-Saban and Chadeaux all participated in this discussion, each presenting their own laboratory's techniques regarding cystine measurement. Topics

covered included the pitfalls of sample preparation, and differing end results in relation to actual transport media and storage. It was agreed that a standard protocol should be agreed upon, in order to avoid discrepancies in future laboratory results.

Dr. William vant'Hoff spoke at length on cystine levels in clinical practice. In terms of knowing what level to aim for, he suggested a figure of $<1\text{nmol } 1/2$ cystine/mg of protein as being the upper limit for heterozygotes with no pathological effects of moderate cystine accumulation. He stated that there is, however, no ideal or safe cystine level, and that leucocyte cystine levels should be monitored 3 monthly.

Dr. Mick Henderson completed the day's presentations with a brief overview of all significant variables, questioning whether or not quality control of the process as a whole was possible, taking into account transport of blood, isolation of cells, and cystine and protein assaying. It was established that this is indeed a very difficult thing to standardise, although quality control of the assays is relatively straightforward, and should not be overlooked.

Dr. Fowler headed a final discussion, which summarised the day's topics, and encouraged all members to agree on certain standardised guidelines. The final outcome was the decision that guidelines would be drawn up for

the treatment of the disease, and that the group will also prepare guidelines for the preparation of total leucocytes and granulocytes, for sampling and for storage. These guidelines have been distributed to each laboratory performing this measurement. All laboratories involved in this analysis should also be encouraged to use some form of internal quality control system for assays. Finally, an external quality control scheme will be established by ERNDIM, and distributed to all laboratories performing this measurement. This should be implemented from the beginning of next year.

Laboratory guidelines have been drawn up. These have been published in full in the Spring edition of the BIMDG Bulletin, available from the editor, Mick Henderson.

The day proved extremely productive, with enthusiastic support from each member present, and a general consensus was reached to endeavour to standardise laboratory methods. It was also agreed to meet again in the future to keep up to date with progress.

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Secretary's column

Council has met twice this year in February and June. Important progress is being made in achieving recognition of metabolic medicine as a training speciality in medicine as Prof Leonard explains on page 3 of this Newsletter. It is also recognised that similar progress needs to be made in the specialist training of dieticians and laboratory scientists.

I would like to remind those of you attending this years annual meeting at Prague that the format of the SSIEM awards will be different this year. There will still be two awards, one for the best oral submission the other for the best poster. But the awards will now be made at the end of the programme in the same year. Since all the accepted abstracts are published in a special edition of the JIMD it has been decided that short communications from the meeting will not need to be published as a separate feature. So, be warned, don't invest any time preparing them!

Hope to see in Prague,

Kind Regards,

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Chairman's Comments

Interest in biochemical genetics has never been greater. This is exemplified by the increasing number of delegates at national and international meetings, the traffic through metab-I and the fact that membership of the SSIEM has now reached over 1000. (If we knew who the one thousandth member is we might have been able to offer him or her a special gift - maybe a copy of the JIMD with signed photographs of the editors.) However despite this interest there is crisis in the number of clinicians who will be available to provide specialist care in the future. In North America this has been a concern raised at the SIMD. In the UK we had, until recently two dedicated training posts but changes to the allocation of paediatric training numbers place at least one of these at

risk. With the required expansion in specialist numbers and planned retirements it is difficult to feel confident as to how the service needs will be met. Recognition of inherited metabolic disease within the European Union (see the article by Professor James Leonard in this newsletter) will raise the profile of the speciality and may help but we seem to be in for some lean times ahead. The situation for specialist laboratories is also difficult, particularly so when under resourced (as is always the case) and when key personnel become unavailable. We need to develop strategies, national and international, to address these issues. How far the SSIEM should become involved in training and governance issues remains open to debate, particularly with the advance in many of the national inborn error groups. We can clearly

have an influence and should use this constructively.

Finally on a more serious matter, in Manchester's Trafford Centre I overheard the end of a conversation recently. "And when she came home from holiday she found out one of her best friends had suddenly died. They said it was cot death, you know adult cot death". The conversation reminded me of a certain sketch from Monty Python. Has this anything to do with the SSIEM or biochemical genetics? Probably not, although perhaps the friend suffered from an undiagnosed fat oxidation defect. A tenuous link. Hope to see you all in Prague.

John Walter,
Chairman,
SSIEM Council

Council Members:

Kim Bartlett
Guy Besley
Jane Collins
Mick Henderson
Cornelis Jakobs
Phillip Lee
Graham Shortland
Kurt Ullrich
John Walter

Corresponding Members:

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AUSTRIA	Sylvia Stockler-Ipsiroglu
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SWEDEN	Elisabeth Holme
THE NETHERLANDS	Frans Trijbels
USA	Gerry Berry

MEETINGS DIARY

SECOND WORKSHOP ON TRIMETHYLAMINURIA

SEPTEMBER 21 - 22, 2001

NIH, BETHESDA

FURTHER INFORMATION:

[HTTP://RARE DISEASES.INFO.NIH.GOV/ORD/NEWS-REPORTS/CALENDAR.HTML](http://rarediseases.info.nih.gov/ord/news-reports/calendar.html)

ITALIAN SOCIETY FOR THE STUDY OF INHERITED METABOLIC DISEASES (SISMME)

ITALIAN SOCIETY FOR NEONATAL SCREEING

WORKING GROUP ON CLINICAL GENETICS OF THE ITALIAN SOCIETY OF PEDIATRICS

8-10TH NOVEMBER, 2001

NAPLES

FURTHER INFORMATION:

[WWW.SISMME.IT](http://www.sismme.it) (NEWS PAGE); [HTTP://MCWEB.UNICA.IT/SISN/](http://mcweb.unica.it/sisn/)

BRITISH INHERITED METABOLIC DISEASE GROUP

WORKSHOP: ORGANIC ACIDURIAS

FRIDAY 16TH NOVEMBER 2001

THE CHILDREN'S HOSPITAL, TEMPLE STREET, DUBLIN

FURTHER INFORMATION:

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5TH MEETING OF THE INTERNATIONAL SOCIETY FOR NEONATAL SCREENING (ISNS)

26 - 29TH JUNE 2002

GENOVA

FURTHER INFORMATION:

[WWW.ISNS-NEOSCREENING.ORG](http://www.isns-neoscreening.org)

RECOGNITION OF PAEDIATRIC METABOLIC MEDICINE IN EUROPE

An application has been made to the European Board of Paediatrics (EBP) for the recognition of Paediatric Metabolic Medicine as a sub-speciality. The Board agreed that the application should be submitted to Committee of European Specialists in Paediatrics (CESP), subject to some minor modifications. CESP also approved the application provided the modifications were agreed. The next step is for the submission to go to Union of European Medical Specialists (UEMS) in March 2002. No major problems are anticipated.

The implications of the recognition are potentially far reaching. Ultimately only those recognised as specialists in the field may be referred patients with metabolic disorders but that is far into the future. The immediate situation once the application has been approved is:

1. If a country wishes to recognise specialists in the field it will have to adopt criteria that are consistent with those in the application. This document will be available once it has been approved.
2. If a country wishes to train

specialists in paediatric metabolic medicine a training programme will have to be established that also meets the criteria laid out in the application. This will be the responsibility of individual countries. This is necessary because the definition and training of paediatricians varies widely throughout Europe.

3. The SSIEM will have to establish a training committee to ensure that the training centres meet the agreed criteria and that the training programme covers the syllabus (previously circulated) satisfactorily. However in the first instance the committee will collect information and only later will inspect individual centres.

Once the application has finally been approved requests for information will be circulated. Even at this stage it would be very helpful if members in European countries would notify Professor Leonard, Chairman-designate of the training committee of the details of any plans.

A. Is it likely that the sub-speciality will be recognised in your country?

B. A nominated representative in the country.

C. The national body that is responsible for establishing and monitoring training programmes.

D. Possible training centres

E. A brief outline of the training programmes.

It is anticipated that not all countries will participate, although there may be advantages of so doing. It may help to obtain resources and finance. We would also hope that it would be easier for those who wish to study at other centres (within their own country and abroad) to obtain the necessary funding but, of course, this is not certain.

Prof J V Leonard

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