

Newsletter Spring 2015

Message from the Chairman

Dear Colleagues

ERNDIM continues to thrive and I would like to thank you all for your support. 2014 saw a continued growth of participations and the numbers of laboratories participating in our EQA schemes. There were 374 laboratories participating from 59 countries. We also continue to gradually expand the scope of what we do and we had three pilot schemes running last year, see page 3 for an update on these.

We are very much hoping that 2015 will be a significant year for ERNDIM. We are preparing to apply for accreditation under ISO 17043 through the offices of the UK Accreditation Services, UKAS. We had a preliminary face to face meeting with inspectors from UKAS last Autumn and have registered ERNDIM as an 'interested party' for accreditation. Following an overview of our activities and organisational structure the inspectors encouraged us to work towards applying for an initial inspection later in 2015. So, much effort is now being put into the creation of a Quality Manual and the supporting documentation. Follow our progress on the website. I would like to pay tribute to, and personally thank, Sara Gardner who has taken the lead on most of this work.

We are planning to open a new DPT EQA group for the Pacific Rim countries. This will be based in Australia and run by Veronica Wiley and Kevin Carpenter. Jim Bonham has written a useful and interesting article explaining the background to this and giving a little history lesson about ERNDIM. This is included on page 4 of the Newsletter.

And finally I would like to bid you farewell as Chairman. I have completed two terms of office plus an additional year, as agreed with the Board of Trustees, to enable us to get to the point of applying for accreditation. I will remain associated with the Executive to help with the accreditation processes until complete. We have planned the succession. From next January ERNDIM will be chaired by Dr George Ruiter (Rotterdam) and so will be in very safe hands.

Very Best wishes for 2015,

Mick Henderson

On behalf of the ERNDIM Executive Committee

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Progress towards Accreditation

We are continuing to make steady progress towards accreditation with the aim being to submit an initial application later this year.

In the past year we have:

- Begun to harmonise EQA scheme annual reports with the aim that all 2014 annual reports will comply with the requirements of ISO 17043.
- Updated the format for the 2014 Certificates of Participation so that they can be sent electronically to

participants.

- Sent performance support letters to all labs that did not submit or only partially submitted results for the 2013 EQA. This is in line with the new poor performance policy and allows us to offer support to labs who are not fully participating in an EQA scheme.
- Launched on-line registration for all 2015 EQA schemes in September 2014. The Registration

website will also allow labs to update their own contact details, view existing EQA scheme orders and check the payment status of their account.

- Met with UKAS in September to discuss the accreditation process and to ensure the steps we are taking are in line with the requirements of ISO 17043.

"Educational Participation was extended to the Quantitative Schemes for 2015 onwards"

Performance Assessment

Critical Error

As we announced last year, the concept of 'critical error' will be introduced for assessment of the 2014 qualitative schemes. A critical error is an error that would be unacceptable to the majority of labs and would have a serious adverse effect on patient management.

A confirmed critical error will mean automatic classification as a poor performer so the final scoring of all qualitative schemes will be discussed at the Spring meeting of the Scientific Advisory Board (SAB). All proposed critical errors will need to be ratified by the SAB before being confirmed. Participants with a confirmed critical error will be sent a Performance Support letter in the same way as labs that score poorly in a scheme.

Appeals Policy

Appeals against classification as a poor performer due to critical error or score can be made by writing to the Administration office within one month of the date of the Performance Support Letter. The full appeals policy is available from the Administration Office upon request.

Educational Participants

This new class of EQA participation was introduced for the 2014 Qualitative Schemes with the aim of helping laboratories who are developing a new service. Educational Participation is subject to a number of conditions and laboratories wishing to register as an Educational Participant must submit a declaration form confirming that the assay is under development in their

laboratory and that they are not at this time providing a clinical service.

Educational Participants still pay the normal EQA scheme fee and receive performance feedback but poor performance will not be recorded and a certificate of participation will not be issued. After 2 years Educational Participants will be expected to move to full EQA participation.

Due to demand from participants Educational Participation was extended to the Quantitative Schemes for 2015 onwards. However it has been decided that Educational Participation will not be available for the DPT scheme due to it covering a number of different types of analysis which makes managing performance issues more complicated.



"Making plans to ensure the resilience and long term stability of ERNDIM"

Participant Survey 2014

We would like to thank everyone who responded to the Participant Survey in June 2014. We received responses from 201 participants from 188 centres in 49 countries; a very healthy response rate of over 50%!

The results from the annual survey help us to continue to improve the quality and efficiency of the ERNDIM schemes and also the service that we offer you so your input is very important to us.

[A full report on the Survey Results is on the website](#) but

briefly 8 out of 12 of the schemes had the same overall scores as last year. The best score for an individual aspect was for 'Frequency of samples' and the most improved scores of the whole survey were for Lysosomal Enzymes (value for money), Qualitative Organic Acids (website display) and Urine MPS (sample volume).

The responses to the questions which assess the overall performance of ERNDIM were very positive

with the overwhelming majority of respondents rating the quality of services provided by ERNDIM as 'excellent' or 'good', and having 'complete' or 'a lot' of confidence that ERNDIM can deliver the service required by participants.

A number of you suggested a qualitative amino acids scheme under 'suggestions for future schemes' and plans for this scheme are now in development! See '[Future pilot schemes](#)' on page 3 for more details.

Participants' Meeting 2015

As usual this will be a half day event held on the first day of the SSIEM conference. This year that will be Tuesday 1st September in Lyon.

Full details will be circulated nearer the time and will be available from the ERNDIM website. Details of the workshop will also be included in the SSIEM Conference programme.

Participation Costs

As you will have noticed, there has been a small increase in the participation fees for the 2015 EQA schemes. This is partly to meet the increasing costs for running ERNDIM as it currently is.

More importantly though, as ERNDIM is moving towards submitting an accreditation application, the amount of administration work is increasing substantially. This, and the administration support that ERNDIM provides for the SSIEM Academy means that we need more clerical support in the Administration office.

We also need to make plans to ensure the resilience and long term stability of ERNDIM and by increasing the fees we will be able to finance a very much needed second scientific administrator in three years' time.

Pilot Schemes

Three new pilot schemes began in 2014: **CSF neurotransmitters** (Prof Simon Heales, London); **Pterins in urine and pterins/DHPR in DBS** (Prof Nenad Blau, Heidelberg); and **Lysosomal Enzymes in DBS** (Dr Kees Schoonderwoerd, Rotterdam).

Regrettably, due to difficulties with sample supply, it is not possible to run the Lysosomal Enzymes in DBS pilot in 2015. However both the CSF neurotransmitters and the Pterins in urine and pterins/DHPR in DBS pilots will run in 2015 and the samples have been dispatched to the participating laboratories. Unfortunately the number of participants in these schemes is limited and registration for these pilots is now closed but we

will publicise any future developments and inform all ERNDIM participants if wider participation is possible.

Future pilot schemes

The current quantitative amino acid scheme adequately tests analytical capabilities but lacks an interpretation component. To overcome this gap we plan to introduce an interpretive (cognitive) amino acid scheme.

Such a scheme has previously ran in the UK and comprised sets of amino acid results together with brief clinical information. Participants interpret the data as if from a patient and submit the results via a web based report.

For the first phase selected labs from the UK, Southern

Germany, Austria and Switzerland will be invited to participate. Scoring will be similar to that used in existing ERNDIM schemes and there will be four assessors, Mary Anne Preece (Birmingham), Rachel Carling (London), Sabine Scholl-Bürgi (Innsbruck) and Brian Fowler (Basel).

Once the structure and logistics have been finalised we will contact the labs selected for the pilot phase with details of how to join the scheme. This will hopefully be in the second half of this year. As with all our pilot schemes participation is limited but we will inform all ERNDIM participants when wider participation is possible.



"We plan to introduce a cognitive amino acids scheme which will focus on interpretation of data"

Training Support Grants

As part of our aim to help improve standards in biochemical genetic testing ERNDIM offers a small number of Training Support Grants each year.

This grant is designed for trainees, in a permanent laboratory position, to gain experience and knowledge in a European ERNDIM approved laboratory in order to develop or introduce new methods to their own laboratory.

Funds can be applied for to cover the travel and accommodation costs incurred by such visits and a maximum of 6 grants will be awarded each year, subject to the approval of the ERNDIM Executive Committee. Full application criteria are given in the application form which can be found on the ERNDIM website under [Training\Grants](#).

In 2014 Ms Inga Baškurova, a Clinical Chemist from Vilnius University Hospital in Lithuania, was awarded a Training Support Grant and in November she visited the Laboratory for Biochemical Genetics in Maastricht University Medical Centre for two weeks.

Inga's laboratory in Vilnius is implementing tandem MS for clinical diagnoses so her time in Maastricht with colleagues who use this technology on a routine basis was extremely helpful.

You can read Inga's full report on her visit on the website under [Training\Travel Grant Reports](#).

Further information on how to apply for an ERNDIM Training grant to support educational visits is on the website under [Training\Grants](#).

Laboratory Directory



In last year's Newsletter we asked laboratories to update their entries in the [ERNDIM Laboratory Directory](#) and invited new labs to join.

There was a good response from Italy, thanks to efforts by Ubaldo Caruso with updates and three new labs joining the directory. Unfortunately the response from other countries was poor. So we can assess future needs for the directory we will use a questionnaire to contact current labs directly as well as obtaining an overview from each country through the National Representatives.

Please contact Brian.Fowler@ukbb.ch for questions about specific issues or general opinions on the directory.



A new DPT Scheme centre for South Asia/Pacific Rim

Jim Bonham, Scientific Advisor, DPT Sheffield

"One of the most helpful of the ERNDIM schemes for newer diagnostic services"



"Labs value the opportunity to discuss and compare their own experience with a defined peer group,"



We are pleased to announce plans for a new centre for the Diagnostic Proficiency Testing (DPT) scheme for South Asia and the Pacific Rim.

The generous offer of support from Veronica Wiley and Kevin Carpenter (both from Sydney, Australia), who will collaborate in the organisation of the new DPT centre, will allow us to launch the new DPT scheme for South Asia and the Pacific Rim from 2016 onwards.

The DPT schemes

This is perhaps one of the most helpful of the ERNDIM schemes for newer diagnostic services: a panel of "real patient" urine samples are provided each year and participants are invited to choose and perform the relevant investigation to establish a diagnosis.

Participating laboratories often comment that the experience offered by access to patient samples with well characterised disorders is very valuable. In particular, the opportunity to discuss and compare their own experience with a defined peer group, typically of around 20 similar labs is valued. This is enhanced if members of staff representing each laboratory can meet face to face.

We are conscious that, while the DPT scheme currently has participants from Pacific Rim countries, the existing DPT organising centres (Sheffield, Basel, Lyon, Amsterdam and Prague) are all within the EU. We have thought for some time that it would be good to develop organising centres for DPT schemes in other places that could be attuned to regional needs and provide a basis for an educational community.

Scheme Organisation

The structure of the new scheme will be modelled on the existing five DPT schemes which now share a common format and results submission website. While the samples distributed differ

Rare Disorders

Rare disorders can be defined, within the EU at least, as those conditions that occur with a prevalence of <1:2,000. While individually rare, collectively they are common affecting one in seventeen people in Europe or around 40 million individuals. 80% of these are genetic in origin and within that group the inherited metabolic disorders (IMD) are an important component.

The experiences facing individuals with rare disorders, while varying person to person, are often characterised by delays and sadly sometimes missed diagnoses, it is reported that one in five people have to wait five years to receive a diagnosis and one in ten people more than ten years.

ERNDIM

ERNDIM was established in 1991 with the aims of:

- Improving diagnosis
- The organisation of laboratory EQA on an international basis
- Acting as an aid to research by allowing greater comparability of results
- Encouraging the adoption of standardised analytical procedures

The EQA schemes offered were initially limited to urinary Qualitative Organic Acids, Quantitative Amino Acids and a single DPT scheme.

The number of participating laboratories have increased (from 138 participants, from 22 countries, in 1995 to 373, from 57 countries, in 2015) as have the number of EQA schemes we offer: we now organise 18 EQA schemes including five DPT schemes which have participants from 30 different countries.

Expansion of IMD services

The growth of ERNDIM has also been paralleled by a growth in the number of countries offering services for the detection and treatment of inherited metabolic disorders over the last twenty years. The development of services in India, China, Indonesia and other Pacific Rim countries has grown and developed along with a rapid growth in services within South America.

Very often these services are pioneered by enthusiastic individuals with a passion for the subject and the benefits that diagnosis can bring to families. These pioneers are often keen to access resources and help from centres in Europe, the US and Australasia. Indeed it is noteworthy that the number of Chinese visitors to the MetBioNet website in 2014 (a UK site offering resources for IMD diagnostic labs, www.metbio.net) was almost half of that from the UK itself.

from centre to centre because of the limitations of sample volume, there is usually one sample which is common to all centres and participants, and the general philosophy of the scheme, operational structure, reporting and assessment are aligned to a single model.

Initially, we will allocate participants to the Sydney centre on a roughly geographic basis but with

space to allow the addition of other participants from within the Region.

It is a big step for ERNDIM to expand outside Europe but it reflects needs within the IMD community as our field develops. Once again we thank the existing scheme organisers for their hard work, provided without individual financial reward, and extend a warm welcome to Veronica and Kevin as they join the team.



ERNDIM collaborated with ETAC to organise the two day training meeting that was held in Paris on 1st and 2nd April 2014. The course was heavily oversubscribed; 108 applications were received and 40 scientists and 44 clinicians attended. The topic of the Academy was storage and neurological disorders. The joint clinical and lab workshops proved very popular and the feedback was positive with all 77 returned evaluation forms agreeing or strongly agreeing that "The programme was relevant to current practice" and 99% agreeing or strongly agreeing that "The academy was relevant to my educational needs".



The topic for the 2015 Academy (20th- 21st April 2015, London) will be congenital Disorders of Glycosylation, Glycogen Storage Disorders, Mitochondrial Diseases and Neurotransmitters.

Registration was in November 2014 and all available places were filled as 109 applications were received for the 80 places.

Information on future Academies will be posted on the ERNDIM website under [News & Events](#).

ESHG GSQC & EuroGentest

Following the completion of the EuroGentest project in January 2014, the ESHG has incorporated EuroGentest in a new structure. The EuroGentest committee will be responsible directly to the ESHG and the ESHG Genetic Services Quality Committee becomes one of 4 sub-committees of EuroGentest: 1) Quality Committee; 2) Professional guidelines, 3) Training, 4) Dissemination.

The new Quality Committee will be streamlined with proposed members to include representatives of EQA providers for cytogenetics, molecular genetics and biochemical genetics alongside representatives of clinical and molecular genetics and expertise in accreditation. Each of these sub-committees will create



working groups to undertake specific tasks and will be financed for the first two years by the ESHG.

Our continuing involvement with the ESHG in this new structure remains important in that: it provides a forum for exchange with our colleagues in other genetic disciplines in striving to improve quality of genetic testing; it provides governance for our activities; it is the final level of appeal in decisions regarding performance attainment for participants of our schemes.

CSCQ Results website

The CSCQ Results website continues to work well. Both the DPT and Urine MPS schemes now use the CSCQ Results website for online submission of results.

This year we are also hoping to move the CDG scheme to the CSCQ results website and our long term plan is that the other qualitative schemes will also use this facility in the future.

If you have any queries or concerns please contact CSCQ;
Xavier.albe@hcuge.ch

ERNDIM Management Committees

Board of Trustees

For the first time the Board meeting in 2014 was chaired by the Chair of the Board of Trustees, Prof Katrin Őunap. Prof Őunap is not a member of the Executive Committee or SAB and provides objective governance for ERNDIM.

Full details of the members of the Board of Trustees, the Executive Committee and the Scientific Advisory Board are on the website ([About Organisation and Key Persons](#)).

ERNDIM Officers
Chair of the Executive Committee:
Mick Henderson,
Leeds, UK

Treasurer:
Jörgen Bierau
Maastricht,
The Netherlands

Secretary:
Viktor Kořich,
Prague
The Czech Republic

Chair of the Scientific Advisory Board:
Christine Vianey-Saban,

Website update (www.erndim.org)

Documents added in 2014

- [Newsletters](#)
 - Newsletter 2015
- Meetings & Reports
- [Meetings](#)
 - Speakers presentations,

ERNDIM meeting,
Innsbruck, Sept. 2014

- Poster presentations by EQA scheme Scientific Advisors, SSIEM, Innsbruck, Sept. 2014

- [Reports](#)
 - Report on the 2014 Participant Survey
 - EQA scheme annual

reports

Training & Education

- [Educational Documents](#)
 - Updated Educational Participant Applicant Declaration Form
- [Travel Grant Reports](#)
 - Travel Grant Report by Inga Bařkirova

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