



Newsletter Spring 2014

Message from the Chairman

Dear Colleagues

We continue to be successful as an organisation seeing increases both in our numbers of participants and our activities. Last year we had 362 participants (an increase of 6% compared to 2012) from 58 countries and we continue to collaborate with the SSIEM to provide training at the annual ETAC Academy. We also like to support educational visits to centres of expertise and set aside a small grant fund each year for that purpose. An example of how this can be of benefit to an individual and their country is illustrated by the short account in this Newsletter of Dr Kali Makedou's visit to Leeds in October 2013.

We have been working to change the composition of the Board of Trustees to make it more independent from the operational management of ERNDIM. We have nearly achieved this goal now. We delighted that Prof Katrin Öunap has agreed to become Chair of the Board from January 2014. This means that the Board is now chaired by someone who is not part of the Executive Committee or the Scientific Advisory Board. And so is able to take a more critical and independent view on the governance of our organisation. A brief biopic about Katrin appears later in the Newsletter.

Very Best wishes for 2014,

Mick Henderson

On behalf of the ERNDIM Executive Committee

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Progress to Accreditation

A key goal for ERNDIM is to become an accredited EQA provider. We intend to apply for accreditation through UKAS, the UK Accreditation Service. We are making steady progress and in the past year we have:

- Appointed an independent Chair for the Board of Trustees.
- Agreed job descriptions for key roles.
- Agreed policies on poor performance, appeals, critical error and educational participants.
- Implemented a formal document control system.

We've also welcomed Alice Brockway as the part-time ERNDIM Administration Assistant. Alice joined us in February last year and has been a huge help dealing with the administration of the EQA schemes.

Performance Assessment

Critical Error

The concept of 'critical error' will be introduced to the assessment of the qualitative schemes this year. 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.

The introduction of critical error is on the advice of the Genetic Services Quality Committee (GSQC) of the European Society of Human Genetics (ESHG), which wants to see harmonisation across all European genetic EQA providers.

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.

Appeals Policy

Alongside the introduction of critical error we have also formalised an appeals policy.

The full details of how to appeal against classification as a poor performer will be available on the website in April but briefly appeals must be submitted in writing to the Administration office within 3 weeks of the date of the Performance Support Letter.

Educational Participants

We have noticed that some laboratories subscribe to EQA schemes but do not submit any results. We know these laboratories may be using the EQA samples for educational purposes so we have introduced a new class of EQA participation for the 2014 Qualitative Schemes, (Continued on page 2)

"Helping laboratories who are developing a new service"



"A key goal for ERNDIM is to become an accredited EQA provider"



Performance Assessment *(Continued from page 1)*

called Educational Participation, with the aim of helping laboratories who are developing a new service. Educational Participation is subject to a number of conditions and due to limited availability of EQA materials the number of Educational participations we can accept per EQA scheme are limited. Laboratories wishing to register as an Educational Participant in a qualitative scheme for 2014 were asked to submit a declaration form last December which confirmed 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.

If the introduction of Educational Participations for the qualitative schemes is successful we hope to extend the availability to the quantitative schemes for the 2015 scheme registrations.

CSCQ Results website for DPT & Urine MPS schemes

The online submission of the DPT results using the CSCQ results website is working very well.

It is much easier for Scientific Advisors to evaluate performance when participants' submissions are structured in a uniform way and feedback from the scheme participants has been very positive.

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

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

Participation Costs

Participation costs have been increased by 3% for all schemes for 2014 to cover the increased costs of running ERNDIM on a more professional basis.

The exception is the Lysosomal Enzymes scheme where the scheme cost has increased from €450 in 2013 to €500 in 2014.

This reflects the greater cost of using fibroblasts as the base material. Fibroblasts give much more realistic enzyme values than the EBV cell lines which were used in 2011.

Fibroblasts were introduced as the base material in 2012 but the increased cost was subsidised by ERNDIM in 2012 and 2013.

Participant Survey 2013

We would like to thank everyone who responded to the Participant Survey in June 2013.

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.

In the 2013 survey the scores for 11 out of 12 of the schemes improved or were the same as in 2012.

The best scoring scheme was Cystine in WBC while the worst scoring schemes were Lysosomal Enzymes and CDG, although the score for Lysosomal Enzymes has improved since 2012.

The best score for individual aspects was for 'Usefulness of the Annual Report' and the most improved score of the whole survey was for Acyl carnitines for 'Adequacy of the report'.

A new set of questions were added to the 2013 survey with the aim of assessing the perception of the overall performance of ERNDIM. The responses to these questions 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 full report on the Survey Results is on the website.](#)

Website update (www.erndim.org)

Documents added in 2013

[Newsletters](#)

- Newsletter 2014

Meetings & Reports

• [Meetings](#)

- ERNDIM speakers presentations, EuroGentest/ESHG Workshop, Paris, June 2013 *(details on page 3)*
- Speakers presentations, ERNDIM meeting, Barcelona, Sept. 2013
- Poster presentations by

EQA scheme Scientific Advisors, ICIEM, Barcelona, Sept. 2013

• [Reports](#)

- Report on the 2013 Participant Survey

Training & Education

• [Educational Documents](#)

- Educational Participant Applicant Declaration Form

• [Travel Grant Reports](#)

- Travel Grant Report by Dr K Makedou

Participants' Meeting 2014

As usual this will be a half day event held on the first day of the SSIEM conference. This year that will be Tuesday 2nd September in Innsbruck.

Full details will be circulated nearer the time and will also be available from the website.

Laboratory Directory



The [ERNDIM Laboratory Directory](#) holds details of laboratories that perform diagnostic tests in Inherited Metabolic Diseases with the aim of aiding specialist workers in the field to select laboratories for sample referral.

We're asking laboratories that are already listed in the ERNDIM Laboratory Directory to update their details and inviting new laboratories to join the Directory.

These can both be done very easily by following the instructions on the right. Instructions for how to view the information already held in the Directory are also shown.

The Directory was developed in the early 2000s and refined as part of the EuroGentest (EUGT) project from 2005 to 2007.

We'd now like to update the information held for participating laboratories and also to re-evaluate whether the Laboratory Directory is needed and fit for purpose so if you think this is still a useful and needed service please email Brian.Fowler@ukbb.ch and let us know.

To access the Laboratory Directory click the 'Laboratory Directory' tab in the ERNDIM website (www.erndim.org) and click on 'Go to the directory' to open the Directory in a new window. You can then:

- **View laboratory information:** Click on 'Laboratories, then select a

country. Click on an institute name to bring up a list of analytes tested there; clicking on 'More info' will bring up the full laboratory listing.

- **Update listed laboratory data:** Click on 'Participants', then on 'Existing Participants'. Select your lab from the list and enter your password. If you have forgotten your password please click on the 'forgot password?' button.

- **Add a new laboratory to the Directory:** Click on 'Participants', then on 'New Participants', add the details for your laboratory and then click on 'register' at the bottom of the page.

- **Applications to add new analytes to the Directory** can be made by emailing Brian.Fowler@ukbb.ch with a justification for the addition.



"The aim of the ERNDIM Directory of laboratories is to aid in selection of laboratories for sample referral"

Pilot Schemes

There are three new pilot schemes in process this year:

- **CSF neurotransmitters:** Scientific Advisor - Prof Simon Heales, London
- **Pterins in urine and pterins/DHPR in DBS:** Scientific Advisor - Prof Nenad Blau, Heidelberg
- **Lysosomal Enzymes in DBS:** Scientific Advisor - Dr Kees Schoonderwoerd, Rotterdam

Registrations for all 3 schemes have taken place and the samples were dispatched to the participating laboratories in mid-February. 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.

EuroGentest/ESHG Workshop:

Interpretation of diagnostic genetic results (biochemical, cytogenetic and molecular)



ERNDIM has been a partner in the Eurogentest project for eight years and an emerging issue has been how to improve quality in genetic testing as reflected by performance in EQA schemes.

With this in mind a workshop was held as a satellite meeting of the ESHG in Paris on June 7th, 2013 which was attended by approximately 90 participants.

The programme consisted of plenary lectures on interpretation and other problems in genetic testing for each of the three disciplines: biochemical

genetics, cytogenetics and molecular genetics. Breakout groups split by discipline also worked on specific cases.

Mick Henderson and Brian Fowler, representing ERNDIM, gave presentations on Complications in Reporting and Examples of common errors from ERNDIM's DPT scheme (both presentations are available on the ERNDIM website under [Meetings & Reports\Meetings](#)).

The workshop was aimed at laboratories that might have difficulties in interpretation of results as indicated by performance in EQA schemes.

ERNDIM offered travel grants of up to €500 to our members; 10 grants were awarded although just six were taken up.

Following positive feedback from participants, and in line with our major aim of improving quality of biochemical genetic testing, it is hoped to repeat the workshop at the next ESHG meeting which will be in Milan (31st May to 3rd June, 2014).

ERNDIM is hoping to again offer travel grants for this workshop and as soon further information is available we will announce the details on our website.

ERNDIM Officers**EC Chair:**

Mick Henderson,
Leeds, UK

Treasurer:

Jörgen Bierau
Maastricht,
The Netherlands

Secretary:

Viktor Kozich,
Prague
The Czech Republic

**Chair of the Scientific
Advisory Board:**

Christine Vianey-Saban,
Lyon, France

Oligosaccharide Kit

An oligosaccharide kit with urine samples representative of the classical oligosaccharidoses is available for purchase through SKML, a partner organisation of ERNDIM's. [Details of how to order the kit are on the website.](#)

A protocol for qualitative TLC oligosaccharide analysis and a picture of a TLC separation of the oligosaccharide kit are both available on the website under [Training\Educational Documents](#).

Dr Kali Makedou makes an Educational visit

Dr Makedou is a laboratory based physician and lecturer in Medical Biochemistry at the University of Thessaloniki, Greece. In October 2013, supported by an ERNDIM grant, she visited the Biochemical Genetics laboratories in Leeds, UK for two weeks.

Kali was particularly keen to learn more about the practicalities of providing a service to diagnose organic acidurias.

This was to build on experience gained at the SSIEM Academy course the previous year as her department wants to set up the assay and provide a service for Northern Greece.

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

Kali's travel and accommodation costs for her visit to Leeds were supported by an ERNDIM Training grant.

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

SSIEM Academy



ERNDIM again collaborated with ETAC to organise the two day training meeting that was held in Lyon on 16th and 17th April 2013. The course was heavily oversubscribed; 111 applications were received and 42 scientists and 40 clinicians attended. The topic of the Academy was organic acids and hyperammonaemia including fatty acid oxidation defects. The joint clinical and lab workshops proved very popular and the feedback was positive with all 74 returned evaluation forms agreeing or strongly agreeing that "The programme was relevant to

current practice" and 98.7% agreeing or strongly agreeing that "The academy was relevant to my educational needs".



The topic for the 2014 Academy (1st - 2nd April 2014, Paris) will be storage and neurological disorders. Registration was in December and all available places were filled as 108 applications were received for the 80 places.

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

Changes to Committee Members

Board of Trustees

There were no changes of membership to the Board this year.

The 2013 Board meeting was chaired by the Chair of the Executive Committee, Dr Mick Henderson, as in the past, but this will change at the next meeting!

In line with an increased independent governance role for the Board, the Chair of the Board will no longer be a member of the Executive Committee or SAB and we are delighted to welcome Prof Katrin Õunap as Chair of

the Board from January 2014 (*see below for a brief biog. of Prof Õunap*).

Executive Committee

The Executive Committee said goodbye and thank you to Prof Jim Bonham who has stepped down as Chair of the Scientific Advisory Board after six years in post. He is succeeded by Dr Christine Vianey-Saban.

Full details of Board and Executive membership are on the website ([About\Organisation and Key Persons](#)).

Biography: Prof Katrin Õunap

Prof Katrin Õunap is Head of the Department of Genetics at Tartu University Hospital, Estonia where she also holds the Chair in Clinical Genetics.

She qualified as a paediatrician in 1988 and began her career in the Medical Genetic Centre at Tartu's Children's Hospital. During her early years she was instrumental in the introduction of newborn screening to Estonia in 1993. She completed her PhD thesis on phenylketonuria in 1999.

Throughout her career Katrin has worked consistently to institute and improve clinical and laboratory services for the diagnosis and treatment of metabolic disorders.

Katrin was invited to join the ERNDIM Board of Trustees in 2009 and now we are delighted to be able to announce that from 1st January 2014 she will take on the role of Chair of the Board.

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