



QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

Newsletter Spring 2013

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## 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 342 participants from 54 countries. And for 2013 there are so far 355 laboratories registered from 58 countries with new countries including Iceland, Pakistan, Russia and the Sultanate of Oman. We are also expanding the schemes offered, see Pilot Schemes (page 2). We continue to collaborate with the SSIEM to provide training at the ETAC Academy (page 4).

Last year we made significant progress with improving and modernising the governance structures of the Foundation. We have redefined the inter relationships of the key committees and specified terms of reference for each. These are posted on the website under 'About us'. We are changing the composition of the Board of Governors to increase its independence from the operations management.

Important announcements about performance assessment follow, read on.....

Very Best wishes for 2013

*Mick Henderson*

On behalf of the ERNDIM Executive Committee

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## Performance Assessment

We are being encouraged by the European Society of Human Genetics to harmonise our scheme performance assessments with the other European genetic laboratory EQA providers. The principal points of difference lie with the allocation of points in the scoring systems and the adoption of the concept of 'critical errors'. Full details of the policies outlined here will be published later in the year.

### Changes to DPT scoring

At a special meeting of the

Scientific Advisory Board in November we agreed that from 2013 the qualitative schemes will all adopt the same scoring system of two points for analytical results and two points for interpretation.

For the DPT scheme, there will no longer be an additional point for suggested further actions or testing. This will be included as part of the interpretation. This will also reduce the total score achievable to twenty four (with six samples) and acceptable performance

will generally be held to be a score above fourteen.

### Critical Error

We are now considering the definition of 'critical error' and will inform all participants how this will affect the scoring. In principle this is a category of error that would be unacceptable to the majority of labs and would have a serious adverse effect on patient management. We are hoping to be able to institute this for 2014.

*(Continued on page 2)*

“A better defined appeals policy”



## Performance Assessment *(Continued from page 1)*

### Appeals Policy

We are also introducing a better defined appeals system. This includes the facility for an ultimate appeal to the ESHG in cases of dispute. We realise how damaging it could be for a participant to be categorised as a poor performer. But ultimately the prime concern must be the best interests of the patients.

### Educational Participants

One of the key aims of ERNDIM is education and to help laboratories provide better services. To help further this aim

we intend to define a new category of participation as ‘educational’. This facility should be available for the participation registrations for 2014.

This category is intended for laboratories who are developing a test and need ERNDIM samples and feedback to facilitate, but who are not offering a clinical service at that time. An educational participation will mean that results are returned but not scored. No certificate of participation will be issued, but neither will poor performance be recorded.

## DPT-CSCQ Results website

The system for entry of the DPT results by the website went live last year and is working well. It is now much easier for scheme advisors to evaluate performance when participants’ replies are structured in a uniform way.

If you have any queries or concerns please contact CSCQ;  
[xavier.albe@hcuge.ch](mailto:xavier.albe@hcuge.ch)

## Oligosaccharide Kit

Dr George Ruijter, Scientific Advisor to the Urine Mucopolysaccharide scheme, has collaborated with Dr Cas Weykamp of SKML, to produce a kit with urine samples representative of the classical oligosaccharidoses. The kits will be able to be purchased via the website later this year. The website will also have pdf images of the TLC patterns that should be obtained.

“Pilot schemes planned for CSF Neurotransmitters and Urine & Blood spot Pterins”



## Pilot Schemes

Proposals are at an advanced stage to develop and pilot two new EQA schemes.

- **CSF neurotransmitters**  
 Scientific advisor: Prof Simon Heales, London

- **Urine and bloodspot pterins**  
 Scientific advisor: Prof Nenad Blau, Heidelberg

Participants were surveyed last year and there were sufficient expressions of interest to make both schemes viable. Additional participants should contact [admin@erndim.org](mailto:admin@erndim.org)

## 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:

- Improved the governance of our committees
- Appointed a Quality Manager
- Strengthened the

oversight function of the Board of Trustees

- Made the DPT schemes more uniform by co-ordinating them through CSCQ
- Begun to write job descriptions for our officers
- Approved funding for a part time office assistant to support the ERNDIM Administrator.

## Participants’ Meeting 2013

This will be a half day event held on the first day of the ICIEM conference. This year that will be Tuesday 3rd September in Barcelona.

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

## Participant Survey 2012

We would like to thank everyone who responded to the Participant Survey in August 2012. 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 2012 we had a total of 137 responses and the overall score for all aspects of all schemes improved when compared to 2011.

The scores for all of the individual schemes improved (except for Cystine in WBC which stayed the same) with the scores for CDG and Quantitative Organic Acids showing the most improvement. The best scoring schemes were Special Assays Urine and Cystine in WBC while the worst scoring schemes were Lysosomal Enzymes and Acyl carnitines. Of the individual aspects

of the schemes the best score was for 'Frequency of samples' and the worst was for 'Value for Money', although this has improved since 2011. The most improved scores (compared to 2011) were for 'Billing Arrangements' and 'Frequency of Samples'. A full report on the results of the 2012 Participant Survey is on the website.



## Website update ([www.erndim.org](http://www.erndim.org))

### Documents added in 2012

- Speakers' Presentations from the ERNDIM meeting, Birmingham, September 2012 (Meetings and Reports/ Meetings)
- Report on the Participant survey 2012 (Meetings and Reports/ Reports)
- Newsletter 2013

## EuroGentest 2 & the ESHG Genetic Services Quality Committee



Our activities fall within Unit 1 of EUGT2 which is headed by Ros Hastings of CEQA. The aims of our work package are:

- improved organisation and coordination of EQA schemes.
- when possible working towards harmonization with our partners in Molecular Genetics and Cytogenetics.
- accreditation of ERNDIM itself and our individual EQA schemes.
- developing generic criteria for assessing poor performance, especially when it is persistent.
- introducing measures to improve performance including the development of best

practice guidelines.

Activities this year have included a meeting of Unit 1 on 19<sup>th</sup> October 2012 in London and a ERNDIM SAB meeting in London on 28<sup>th</sup> of November 2012. At this meeting we discussed and agreed on how to assess performance in a harmonised way within all our proficiency type schemes including developing the concept of "critical error" (page 1).



ERNDIM continues to be represented in the ESHG Quality Committee, to whom we report on all of our activities, by Viktor Kožch and Brian Fowler.

We have met three times in 2012; Amsterdam (23<sup>rd</sup> March); Nuremburg (25<sup>th</sup> June); and London (23<sup>rd</sup> November).

As well as overseeing our EQA activities an important outcome of this committee is the plan to hold a Training Workshop - satellite meeting at the 2013 ESHG Paris conference. The main focus of this workshop will be to address common errors in all our schemes including ERNDIM ones and how these affect performance. ERNDIM participants are encouraged to attend.

Our participation in these activities continues to keep ERNDIM at the forefront of efforts to improve the quality of genetic testing in Europe.

"Participation in these activities continues to keep ERNDIM at the forefront of efforts to improve the quality of genetic testing in Europe"

**ERNDIM Officers****EC Chair:**

Mick Henderson,  
Leeds, UK

**Treasurer:**

Jörgen Bierau,  
Maastricht,  
The Netherlands

**Secretary:**

Viktor Kožich,  
Prague,  
The Czech Republic

**Chair of Scientific  
Advisory Board:**

Jim Bonham,  
Sheffield, UK

**SSIEM Academy**

ERNDIM collaborated with ETAC, to organise the two day training meeting that was held in Manchester on October 2<sup>nd</sup> and 3<sup>rd</sup> 2012. It

was attended by 39 scientists and 28 clinicians. The topic was amino acids, reflecting the topic of the first Academy in Lisbon in 2008. Following feedback from participants at the 2011 Academy we increased the number of joint sessions for this course. There were an additional two workshops where clinical and scientific convenors collaborated. These proved very successful and we are going to repeat them at the 2013 Academy in Lyon.



The 2013 Academy will be held in Lyon on the 16<sup>th</sup> & 17<sup>th</sup> April 2013. The topic will be disorders of organic acids and fatty acid oxidation.

For programme details see the SSIEM website.

**Changes to the Board of Trustees & the Executive Committee****Board of Trustees**

We welcomed three new members elected to the Board in September.

- Mrs Marjolein D. van der Burgt-Spaapen, scientific & communications officer for VKS, the Dutch patient organisation for adults and children with metabolic disorders.
- Mrs Marketa Lhotakova, Chairperson of the CAVO (Czech Association of Rare Diseases), an umbrella

organization for Czech patient groups.

- Dr Cristiano Rizzo who is a Laboratory Scientist, from the Bambino Gesù Children's Hospital in Rome.

The Board said goodbye and thank you to two members who have stepped down, Dr Ries Duran and Dr Alberto Burlina.

**Executive Committee**

The Executive Committee said goodbye and thank you to Dr Christine Vianey-Saban and Anny Brown. Dr Jörgen Bierau takes over as treasurer and Dr Viktor Kožich takes over as secretary.

Full details of Board and Executive membership are available on the website.

**And finally.....**

We'd like to say a very big thank you to Malcolm Heron who will be retiring at the end of March. Malcolm has been ERNDIM's accountant for over 10 years and has played a huge part in

ERNDIM over that time. We wish Malcolm all the best for the future!

We'd also like to welcome DPA Accounting Limited who have taken over as ERNDIM's accountant. DPA

Accounting will be dealing with invoice payments ([finance@erndim.org](mailto:finance@erndim.org)) but for all other queries please contact the ERNDIM Administration Office ([admin@erndim.org](mailto:admin@erndim.org)).

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