European Research Network for evaluation and improvement of screening, Diagnosis and treatment of Inherited disorders of Metabolism

ERNDIM

QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

SUMMER 2020

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Message from the Chair

Dr George Ruijter, Chair, Executive Committee

Dear Colleagues,

The start of 2020 has been rather bizarre due to the COVID19 pandemic. The alienating pictures of struggling hospital employees and empty streets will not be easily forgotten. Because of the COVID19 restrictions imposed by governments in many countries, laboratory personnel could not travel to work or had to work in shifts, resulting in delays in diagnostic testing.

The ERNDIM Executive Committee and Scientific Advisory Board therefore decided in March to postpone result submission deadlines of all schemes until June.

At the time of writing, June 2020, most countries are out of the lockdown and labs are managing to submit their scheme results. We will of course closely monitor the situation and act accordingly.

In this newsletter we will present an update of current ERNDIM activities. In 2018-2019 ERNDIM has experienced further growth both in our numbers of participants and our activities. Last year we had 1953 scheme registrations from 408 participants in 63 countries (an increase of 32% in 5 years, i.e. compared to 2014). In part, this growth is explained by the introduction of a new scheme, Special Assays in Dry Blood Spots in 2019, which has increased the number of different schemes to 16. However, the number of participating labs has also increased by 9% indicating that an increasing number of labs that are active in diagnostics of inborn errors of metabolism acknowledge the importance of external quality assurance and start to participate in ERNDIM schemes.

In 2020 we will continue to organize the current pilot scheme, cognitive amino acids (see page 3 for a short report on 2018-2019 results). New schemes are operated as pilot schemes for at least 2 years before introduction as a regular scheme. If you have ideas on a potential new pilot scheme, please contact the ERNDIM Admin Office.

Our educational activities include co-organisation, with SSIEM, of the SSIEM Academy, which, unfortunately, had to be cancelled in 2020, and providing grants for both scheme participation and visits of individual scientists to centers of expertise.

New in 2019 were the control materials provided for internal QC (in conjunction with our partner SKML/MCA laboratory; see page 3).

Over the years ERNDIM has transformed into a professional organisation, in which the Admin Office team, Sara, Jenny, Kate and Ismenia, play a vital role in all facets of scheme organization and other activities.

High-quality EQA schemes are only feasible with the input of experts from the field and the Executive Committee gratefully acknowledges our Scientific Advisors for their enthusiastic involvement in scheme organisation.

Finally, I would like to congratulate Prof. Dirk Lefeber, Scientific Advisor of the CDG scheme, with obtaining his professorship at Radboud UMC in Nijmegen.

Best wishes

George Ruijter

On behalf of the ERNDIM Executive Committee

A new EQA Scheme: Special Assays in DBS

Rachel Carling, Scientific Advisor

Following a successful 2 year pilot, the ERNDIM Special Assays in Dried Blood Spots (DBS) scheme is now a full EQA scheme. The scheme includes 8 amino acids (phenylalanine, tyrosine, leucine, isoleucine, valine, allo-isoleucine, methionine and total homocysteine), free carnitine, succinyl acetone and NTBC. Participants are **not** required to return results on all analytes.

The initial pilot data indicated that whilst intra-laboratory reproducibility was acceptable at around 10%, standardisation and recovery of all 11 analytes could be improved: the inter-laboratory CV ranges from 20.3% for tyrosine to 112% for succinylacetone. We hope that the scheme will provide participants with assistance to do this.

Unlike many of the other ERNDIM schemes, laboratories are measuring this panel of analytes by a number of different methods. This makes interpretation of the data for the annual report more challenging and we are investigating ways to improve our approach in the future, starting with the circulation of a detailed methodology questionnaire in 2020.

The recent publication of management guidelines for Phenylketonuria, Homocystinuria and Maple Syrup Urine Disease (MSUD) [1-3] has stimulated renewed interest in the traditional DBS assays (phenylalanine & tyrosine, BCAA). Whilst these tests are commonly used to monitor patients with inherited metabolic disease, they are typically legacy assays, introduced into the clinical laboratory in the late 1990's, and their analytical performance is less than ideal. Conversely, assays for succinyl acetone and NTBC in DBS specimens were introduced more recently and are currently only provided by a small number of participants. Both tests show significant under-recovery and a large inter-laboratory CV.

There is now significant evidence demonstrating how the quality of the DBS specimen can affect the analytical result. The volume of blood applied to the filter paper, spot size and quality, haematocrit and punch location are all critical factors. The introduction of commercially available, DBS collection devices that collect defined volumes of liquid blood and are reported to overcome some of these issues, will also focus attention on DBS testing.

ERNDIM was founded in the early 1990's, not just as a means of providing EQA, but to tackle the issues of comparability of results, clinical guidelines and research findings in rare disorders where conclusions and recommendations made in one centre may not apply universally unless standardisation is in place. As such, we believe the introduction of the ERNDIM DBS scheme is very timely and we look forward to working with participants to improve the quality of these tests.

1. van Spronsen, F.J.; van Wegberg, A.M.; Ahring, K.; et al Key European guidelines for the diagnosis and management of patients with phenylketonuria. *Lancet Diabetes & Endocrinol* **2017**,5, 743-756.
2. Morris, A.A.M.; Kožich, V.; Santra, S. et al. Guidelines for the diagnosis and management of cystathionine beta-synthase deficiency. *J. Inherit. Metab. Dis.* **2017**, 40, 49.
3. Frazier, D.M.; Allgeier, C.; Homer, C.; Marriage, B.J.; Ogata, B.; Rohr, F.; Splett, P.L.; Stembridge, A.; Singh, R.H. Nutrition management guideline for maple syrup urine disease: an evidence- and consensus-based approach. *Mol. Genet. Metab.* **2014**, 112, 210-217.

"...The recent publication of management guidelines for Phenylketonuria, Homocystinuria and Maple Syrup Urine Disease has stimulated renewed interest in the traditional DBS assays schemes..."

EQA Schemes update

EQA Calendar

In 2018 we again made progress towards our aim of publishing the certificates of participation within the first quarter after the end of the scheme year. The 2018 certificates were published in April 2019, earlier than in the 2017 certificates were issued.

Unfortunately, issues due to Covid-19 have delayed the publication of the 2019 certificates so these were not published until June 2020.

In recognition of the issues all our participants have faced due to Covid-19, al the 2020 March, April and May results submission deadlines were delayed to June 2020.

Scoring Policies

The inclusion of a scored interpretative element for the Lysosomal Enzymes in fibroblasts (LEFB), Pterins in Urine, (PTU) and Neurotransmitters in CSF (NCSF) schemes was piloted during 2019. The pilots for the LEFB and NSCF schemes were successful and for 2020 the scoring of interpretation will be part of the performance evaluation for these schemes. The pilot for scored interpretations will continue in 2020 for the PTU scheme and a similar pilot will start in 2020 for the Cystine in WBC scheme.

We will let all participants know in advance of any changes to the scoring policies or the calendar but if you have any questions please email admin@erndim.org.

Progress towards Accreditation

We are still making slow but steady progress towards applying for accreditation. In the past year we have:

- Increased the number of internal audits we are carrying out with the aim covering the whole of the ISO 17043 standard in a 3-4 year cycle.
- Improved the scheme instruction templates so they are now compliant with ISO17043.
- Carried out competence reviews of sub-contractors
- Introduced a new scheme plan template with the aim of introducing this for the 2021 schemes.
- Continued to formalise many of the administration procedures for the Administration Office.
- Continued progress on formalising our relationships with our subcontractors.
- Introduced online results submission for the CDG scheme
- Recruited 2 new part-time Administration Assistants, to replace Alice Brockway and Katie Sheils, who have both moved on to new challenges. We were all extremely happy to welcome Kate Straznikiewicz and Ismenia da Silva to the Admin Office in February 2019 and 2020, respectively.

Internal Quality Control Materials

Many laboratories in the field of screening of IEM struggle with the non-availability of suitable control materials. In the materials that are available analytes are either missing or do not have the required concentration. Home-made controls are an option but it is cumbersome and expensive to obtain all required the analytes. Therefore ERNDIM decided to develop control materials, in collaboration with MCA Laboratories (Netherlands, the scheme organiser for the quantitative ERNDIM EQA schemes) and make them available as an additional service to the users of the ERNDIM external quality control schemes. The range of analytes and their concentrations have been chosen in close collaboration with the respective scientific advisers of the corresponding EQA programmes.

Group Analytes	Content of a package
Amino Acids	6 vials with low concentrations and 6 vials with high concentrations
Organic Acids	
Special Assays in Serum	
Special Assays in Urine	
Acylcarnitines	
Purines & Pyrimidines	
Homocysteine	6 vials with either a low, medium, or high concentration
Pterins	Under development

Figure 1: Available IQ Control materials

Data Management System

In conjunction with the samples, there is also a data management system. The strength of this system is that it does not only monitor the data of the laboratory, but also compares the labs results with labs using the same batch of internal control materials.

More information

Detailed information on the ERNDIM internal control system can be found on: www.ERNDIMQA.nl, under General Information\Control Materials.

Please note all orders and payments for the Control Materials must be made directly with MCA Laboratories and not with ERNDIM.

"…The strength of this system is that it does not only monitor the data of the laboratory, but also compares the labs results with labs using the same batch of internal control materials..."

Pilot Schemes

Cognitive Amino Acids

Brian Fowler, on behalf of co-assessors Mary Anne Preece, Sabine Scholl-Bürgi and Rachel Carling.

A pilot study for this scheme ran from 2017-2019 with 4 circulations, each with 3 sets of sample results. Participation needed to be restricted to not more than about 50 participants especially due to the need to handle data manually using excel tables. Our experience with the pilot was reported at the ERNDIM workshop in Rotterdam, last September by Mary Anne Preece. See a full report which includes a summary of feedback from participants on the ERNDIM website (https://www.erndim.org/store/docs/2019ERNDIMCognitiveAmino-FAGAPAGA961324-24-10-2019.pdf)

At the meeting of the Scientific Advisory Board held in Manchester last November it was decided to move ahead to establish this type of scheme as a permanent ERNDIM activity but this depends on the development of a web based results entry and evaluation programme as well as forming a network of assessors to handle the potentially large number of participants and similar schemes for other analytes.

An initial meeting between one of us and CSCQ has taken place and we hope to see the website programme development as soon possible. In the meantime we plan two more circulations this year in the same way as before.

Brian Fowler, on behalf of co-assessors Mary Anne Preece, Sabine Scholl-Bürgi and Rachel Carling.

Reports for this pilot scheme are on the ERNDIM website under Meetings & Reports\EQA Scheme Annual Reports

Participant Survey [2018 & 2019 scheme years]

We would like to thank everyone who responded to the Participant Surveys in early 2019 and 2020.

In 2020 we received 217 responses from 206 centres in 51 countries, giving a response rate, by centres, of 50%.

The results from this 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.

Full reports on the Surveys Results are on the website but briefly 8 out of 16 of the schemes had the same overall scores as last year with 6 schemes (ACDB, DPT, PTU, QLOU, QTOU and UMPS*) having slightly improved scores. The best scores for an individual aspects were for 'Frequency of samples', 'Adequacy of the report' and 'Usefulness of the annual report'.

The best scores of the whole survey were for 'Frequency of samples' (PTU)*, 'Adequacy of the reports' (DPT)*, 'Website display' (PTU) and 'Usefulness of the annual report' (DPT & PTU).

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.

We also asked questions relating to the possible introduction of pilot schemes for Metabolomics & Lysosphingolipids in the future.

With regards to a Lysosphingolipids pilot 20/206 (9.7%) respondents responded that they would be interested in a pilot, a further 9 (4.4%) responded they would be interested in specific analytes.

With regards to a potential Metabolomics pilot 206 participants responded to 3 questions. The majority of participants (151/206, 73.3%) do not currently have a Metabolomics panel in use or development. The remaining participants have a panel in use (6.3%), development (12.1%) or available for research only (8.3%). The preferred matrix for this pilot is Plasma (43/91, 47.3%).

[*ACDB = Acylcarnitines in DBS, DPT = Diagnostic Proficiency test, PTU = Pterins in urine, QLOU = Qualitative Organic Acids in urine, QTOU = Quantitative Organic Acids in urine, UMPS = Urine Mucopolysaccharides.]]

"Details of the types of samples that are needed can be found on the EQA schemes tab of the ERNDIM website"

Appeal for donated samples

If you would be able to collect a clinical sample that could be used as an EQA sample for one of ERNDIM's qualitative schemes please contact the Administration Office who will send you the ERNDIM consent form and details of how and where to send the samples.

Details of the types of samples that are needed can be found on the EQA schemes tab of the ERNDIM website but please contact admin@erndim.org before sending any samples and please do not send any samples to the Administration Office.

If you have any questions please contact the Administration Office (admin@erndim.org).

Chromatogram Library

This project has remained dormant since the last newsletter. Nevertheless it remains an important aim of ERNDIM to provide what we believe to be a useful resource that will benefit ERNDIM participants. We hope to open up the directory for public use with a limited number of about 60 disorders within the next two months.

Laboratory Directory

Our previous plans, in collaboration with MetabERN, to update the laboratory directory have been reassessed within the context of moves to integrate our "Analyte focused" approach with the "Disease oriented" system that is characteristic of the extensive Orphanet website. Also there is a desire to coordinate laboratory tests with the IEMBase (http://www.iembase.org/) as well as the new International Classification of Inherited Metabolic Disorders developed by Carlos Fereira. We are carefully considering the best way to bring our aims to fruition bearing in mind limited resources and person power.

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 Executive Committee. Full application criteria are given in the application form which can be found on the ERNDIM website under Training\Grants.

Since the last Newsletter, 1 training support grant has been awarded :

- Anna Saada from the Hadassah Medical Center Jerusalem, Israel visited CMMS at the Karolinska University Hospital in Stockholm, Sweden to receive training in techniques important for the diagnosis and treatment of IEM, with the main focus on methodologies related to organic acids, acylcarnitines and amino acids

You can read Anna's reports on her visit and all the reports on previous visits on the website under Training\Travel Grant Reports.

"ERNDIM offers a small number of Training Support Grants each year"

MetabERN; The journey so far

Maurizio Scarpa, Coordinator on behalf of all the MetabERN Members, Udine University Hospital, Italy

Activities and main results

The Clinical/Research Network named "European Reference Network on Rare Hereditary Metabolic Diseases", (acronym "MetabERN"), has been established thanks to the Cross-border Health Care directive and is being funded within the framework of the Third Programme for the Union's action in the field of Health. MetabERN represents the first comprehensive, pan-metabolic, pan-European, patient-orientated platform, aimed to transform how care is provided to patients with inherited metabolic diseases (IMDs) in Europe.

The **main goal of this initiative** is to ensure coordinated action in creating the widest possible collaboration among paediatric and adult metabolic physicians and patient associations at EU level, facilitating patient access to specialists with expertise in the metabolic field and to foster research activity.

The network is currently involving 77 Health Care Providers (HCPs) from 23 European Member States, and aims to reach out and collaborate with all expert centers in EU creating the widest possible collaboration among paediatric and adult metabolic physicians and patient associations at EU level.

MetabERN considers the correct laboratory diagnosis as a key factor for successful management of patients. To this aim a formal collaboration with ERNDIM has been established in 2016 to ensure the best quality of laboratory services among MetabERN members. To do that MetabERN is considering to request mandatory participation of MetabERN laboratories in External Quality Assurance (EQA) schemes provided by ERNDIM to assure the level of adequate performance. The collaboration is also aimed to discuss modification of existing EQA schemes as well as introduction of pilots for new schemes in case there would be such need. In addition, MetabERN has initiated a dialog with ERDNIM to initiate collaboration for improving and maintaining good quality of laboratory services improving, assuring and maintaining an adequate level of education in biochemical genetics for the new generation of laboratory specialists.

In the metabolic field there is a big demand for updated information on diagnosis and diagnostics and in this regard MetabERN would like to make a detailed directory of diagnostic laboratories of high standards in combination with information on all the different inborn errors of metabolism. The idea is to make available a database that could be useful for both health workers on different levels and patients with IMDs.

Orphanet (supported by grants from the European Commission) is an existing database on rare diseases including the IEM. The aims of MetabERN are very similar to those of Orphanet: to provide high-quality information on rare diseases, and ensure equal access to knowledge for all stakeholders.

This is why the idea was born to start the shared initiative by MetabERN, ERNDIM and Orphanet to use and implement the functionalities of existing databases. A cooperation between these organisations supported by MetabERN could be beneficial to all parties and result in a detailed, up-to-date directory which could be a useful tool for doctors as well as patients.

For more information visit http://metab.ern-net.eu/

Workshop on the future of ERNDIM schemes, November 2018

On November 28, 2018, we organised a workshop to discuss future developments in the ERNDIM schemes. Our motivation was the fact that laboratory diagnostics of IEM is subject to change and ERNDIM needs to follow such changes and provide EQA schemes needed by our customers.

Labs sometimes request more samples in the quantitative schemes (i.e. 12 instead of 8) to enable continuous monitoring of quality, but due to logistic issues we concluded that this is currently not feasible. Instead, we will try to implement more equal distribution of submission deadlines in a scheme year.

Some of our proficiency testing (qualitative) schemes have always focused on metabolites groups (organic acids in urine, acylcarnitines in DBS); while in fact we are testing the ability to diagnose disorders. We have already noticed that some participants use novel MS methods and also measure e.g. amino acids in these samples. During the workshop we have discussed the possibility to merge our proficiency testing schemes to focus on different disease groups. We need to investigate such change further and will inform you later if we decide to proceed.

Ideas for new compounds and schemes included (lysosphingolipids, additional compounds in the Special Assays DBS scheme, and targeted and untargeted metabolomics. Please let us know your interest in these ideas by responding to questions on new schemes that we will include in the annual participant survey.

A cognitive scheme, i.e. interpretation of metabolite profiles, is an established method to test proficiency of labs as well as individual laboratory specialists. We intend to introduce more cognitive schemes when the Cognitive Amino Acid pilot scheme is completed and software is available to efficiently run such schemes.

Other possible future activities that we discussed included new educational kits containing positive IEM samples and setting up a guideline for sample exchange or tests to perform in case EQA is not available.

"...laboratory diagnostics of IEM is subject to change and ERNDIM needs to follow such changes and provide EQA schemes needed by our customers."

ERNDIM Participants' Meetings

2019 Meeting, Rotterdam, Netherlands

On 3rd September 2019 the annual ERNDIM participant meeting was held in the De Doelen International Conference Centre in Rotterdam, Netherlands. The meeting was well attended.

As usual, the programme started with the 5 DPT scheme participant workshops.

This was followed by an update from the ERNDIM Chair, George Ruijter and discussion of the 2019 DPT scheme common sample led by Alessio Cremonesi from Zurich.

Mary Anne Preece gave a presentation on the progress to date with the Cognitive Amino Acids pilot scheme while M. Ángeles García Cazorla gave a presentation on Defects of BCAA metabolism.

Presentations from the meeting can be found on the ERNDIM website under Meetings and Reports\Meetings.

2020 Meeting

The annual ERNDIM participant meeting is always organized on the Tuesday morning before the SSIEM symposium. Since this year's SSIEM symposium in Freiburg, Germany, has been canceled due to the COVID19 pandemic, no meeting will be organized in 2020. As an alternative to the DPT workshops, we will organize video conferences to discuss the results of the Diagnostic Proficiency (DPT) schemes with participants.

We will send all participants the details of how to access the video conferences when they have been agreed.

2021 Meeting

In 2021, the international ICIEM conference will be held in Sydney, Australia. As in 2009 and 2017, we will organize a separate ERNDIM symposium in Europe, in the autumn of 2021. The exact dates, location and program will be announced in due time.

CSCQ Results Website

CSCQ are continuing to work on expanding and improving the Results website for the qualitative schemes. We were very pleased that online results submission was made available in 2018 for the Qualitative Organic Acids and Acylcarnitines in DBS schemes and in 2019 for the CDG scheme. This means that online submission is now available for all full ERNDIM EQA schemes!

Testing and development work for evaluation and reporting software modules, which will allow individual lab annual reports to be produced, is still ongoing for many of the qualitative schemes.

We are very grateful for support from SSIEM in funding the development of these CSCQ website extensions which has allowed a more rapid transition to online results submission for these schemes than would otherwise have been possible.

National Representatives

The ERNDIM National Representatives are key members of ERNDIM and assist the ERNDIM Management Committees in disseminating the activities of the network to laboratories in their country. National Representatives are selected by ERNDIM, where possible, following nominations by the National Society.

We remind all ERNDIM participants that your representative is available for questions on all aspects of ERNDIM, especially local issues.

"The ESHG QSC… maintaining and improving the quality of clinical genetics and genetic laboratory services across Europe."

ESHG EuroGentest Quality Subcommittee

Mick Henderson, ERNDIM Quality Lead

What is the function of the ESHG QSC?

I represented ERNDIM at the Quality Sub Committee of EuroGentest for nearly two years now. The first was an overlap year with Brian Fowler who had been our original contact with this group and was a founder member of its parent, EuroGentest. The Quality Sub Committee of EuroGentest (QSC) is itself a subcommittee of The European Society of Human Genetics. The ESHG QSC has a wide remit that can be summarised as maintaining and improving the quality of clinical genetics and genetic laboratory services across Europe. The detailed remit and composition of the group is outlined on the ESHG website: https://www.eshg.org/index.php?id=gsqc

One key function was to monitor performance of participating laboratories in the various genetic laboratory EQA schemes. An annual questionnaire is sent to each EQA provider, including ERNDIM, to collect feedback on the performance data for each EQA scheme. This includes numbers of poor performers, persistent poor performers and any critical errors, The shared data is scheme specific but totally anonymous so no participants are named. The aim is to work towards harmonisation of performance evaluation, and to help less well developed schemes achieve consistent and high standards of governance. It was also motivational in the introduction of the concept of critical error to all the schemes.

Other areas of activity include:

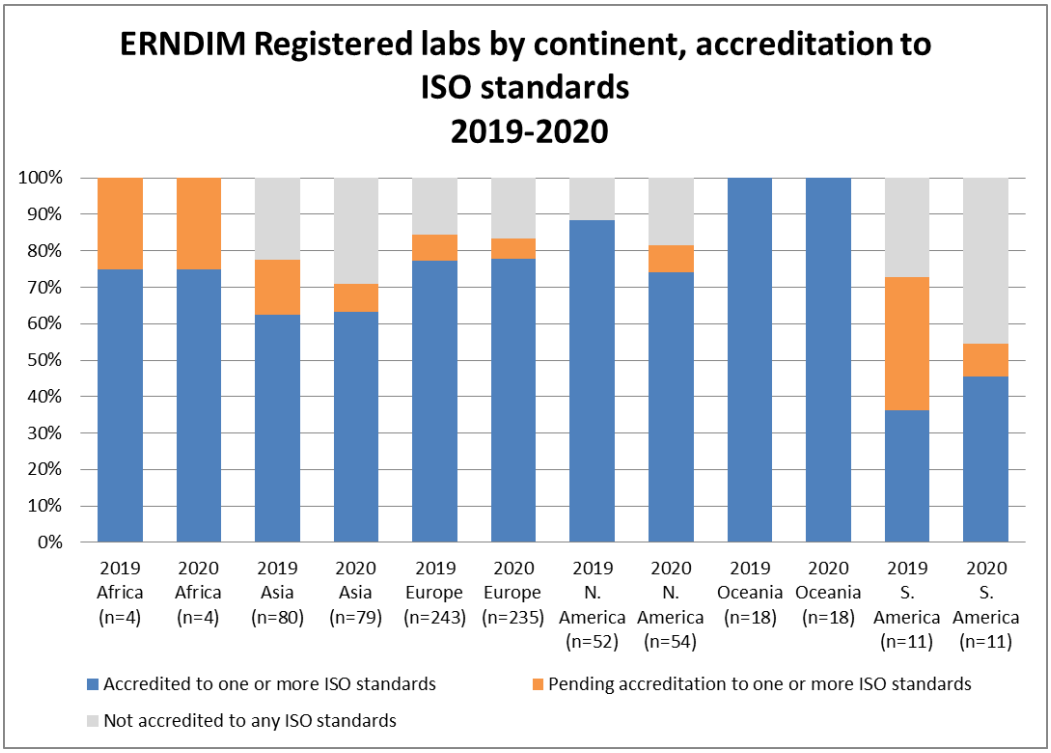
- Planning a Performance Monitoring Workshop for September 2020. This will be by invitation only but will attempt to have representation from all the major stakeholders, i.e. scheme providers, accreditation bodies, national bodies with responsibility for EQA governance oversight. The aim is to produce an advisory report that will underline the importance of EQA and hopefully recommend national governance procedures.
- There is growing concern over the implications for impending EU legislation about IVDs (in vitro diagnostic devices). The anxiety is that there will be pressure on laboratories to change from in-house assays to more expensive CE marked kits, wherever they exist. Uncertainty exists around how laboratories can make acceptable arguments for 'equivalence'. This will, of course, affect not just genetic laboratories but all clinical laboratories. A collaboration, The Biomedical Alliance, has been formed so that a unified voice can put arguments more powerfully to the EU Commission. At present genetic laboratories have not been invited to be part of this Alliance, but the ESHG QSC is now trying to gain membership and hopes to send a representative to their meetings.
- A review and update of the Laboratory Reporting Guidelines, published by members of the ESHG QSC (Claustres et al *Eur J Hum Genet* (2014) 22, 160-170) is planned to take place this year.

I think it's fair to say that ERNDIM makes a valuable contribution to the work of this committee. We have been established for longer than most of the other scheme providers and we are very well organised. But we also learn ourselves and benefit very much from keeping in touch with the leading edge of European EQA governance.

Accreditation Status of ERNDIM Participants

Since 2017, during registration for the EQA schemes, labs are asked to complete a short survey asking for details of the ISO standards they are accredited to. For the 2019 and 2020 EQA schemes, all of the labs that registered (408 and 401 respectively) completed the survey which asked if they were accredited to ISO 9001, ISO 15189, ISO 17025 or another ISO standard. In 2019 309/408 responding labs (76%) replied that they were accredited to one or more ISO standards this decreased by 2% to 299 in 2020. A further 34/408 labs (8%) reported that their accreditation to one or more ISO standards is pending in 2019, this decreased by 3% to 25/401 labs with accreditation pending in 2020. While in 2019 there were 65/408 labs (16%) stating that their lab was not accredited (or had any pending applications) for any of the specified ISO standards, this increased by 18% to 77/401 labs in 2020.

A decrease in the number of registered labs for whom accreditation was pending can be seen from 10% in 2018 to 8% in 2019. and again to 6% in 2020 The number of labs who are not currently registered to any ISO standards and have no accreditation pending has reduced from 18% (71 labs) to 16% (65 labs) in 2019 but rose slightly to 19% (77) in 2020.



"The joint clinical and lab workshops proved very popular and the feedback was overwhelmingly positive".

SSIEM Academy

SSIEM 2019 Academy

ERNDIM collaborated with ETAC to organise the two day training meeting that was held in Winterthur, Switzerland on the 29th and 30th April 2019.

The course was heavily oversubscribed:129 applications were received and 47 scientists and 82 clinicians attended.

The topics of the Academy were mitochondrial diseases, glycogen storage diseases, neurotransmitters and congenital disorders of glycosylation .

The joint clinical and lab workshops proved very popular and the feedback was overwhelmingly positive with over 95% of the returned evaluation forms agreeing or strongly agreeing that "The programme was relevant to current practice" and that "The academy was relevant to my educational needs".

SSIEM 2020 Academy

The 2020 Academy was planned to take place in Amsterdam, Netherlands in April 2019 and the topics were intended to be Aminoacidopathies, Hyperammonaemia and Urea Cycle Defects. Due to restrictions put in place relating to the corona virus pandemic this has been postponed until April 2021, covering the same topics.

ERNDIM Management Committees

Scientific Advisory Board (SAB)

There have been a number of changes to the membership of the SAB for the 2019 & 2020 scheme years:

- ACDB Rome: Dr Sara Boenzi is now deputy Scientific Advisor.
- Acylcarnitines in Serum: Mr Isaac Ferrrer-López is now deputy Scientific Advisor.
- DPT UK: Ms. Claire Hart is now deputy Scientific Advisor.
- Lysosomal Enzymes in Fibroblasts: Ms Marie Jackson (London) is now Scientific Advisor and Dr Derek Burke is the deputy Scientific Advisor.
- Neurotransmitters in CSF: Dr Simon Pope is now the Scientific Advisor with Prof. Simon Heales remaining involved as the deputy Scientific Advisor.
- Special Assays in Serum and Urine (SAS & SAU): Dr Rafael Artuch is now Scientific Advisor with Dr Mercedes Casado as deputy Scientific Advisor.

Executive Committee

Anny Brown took over as Treasurer in 2018, with Jorgen Bierau taking over as Secretary following the retirement of Viktor Kožich.

Board of Trustees

Begoña Merino stepped down as a Trustee at the end of 2018. In 2020 Mick Henderson took over as Chair of the Board after Katrin Õunap completed her second full term in that role at the end of 2019.

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).

Website update (www.erndim.org)

Documents added since the last Newsletter

Newsletters
- Newsletter 2020

EQA Schemes
- 2020 EQA scheme calendar

Meetings
- Presentations, ERNDIM workshops, Sep. 2018 and Sep. 2019.

Reports
- Report on the 2018, 2019 & 2020 Participant Surveys
- 2018 & 2019 EQA scheme annual reports
- Critical Errors in the 2017, 2018 & 2019 Qualitative EQA schemes

Travel Grant Reports
- Travel Grant Reports by Dr Ann Saada (Reisch).

Educational Documents
- Please note that Educational documents are available on the website under the tab "Training": Several have been updated during 2019. The goal of all ERNDIM Scientific Advisors is to review them all and produce updated documents where required. If you are willing to help us, please contact admin@erndim.org.

Registration Website (www.erndim.org/qa)

Under the Participant Information tab which is only accessible if you log into the Registration Website:

• General Information
- ERNDIM Participants' Guide 2020
- ERNDIM Registration Website manual 2020
- Educational Participation Application forms
- Link to Repeat Sample Request form for 2020

• EQA Schemes
- 2020 EQA scheme instructions

ERNDIM
QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

"Working towards a consensus between Biochemical Genetics Centres on reliable and standardised procedures for diagnosis, treatment and monitoring of inherited metabolic diseases"

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