ERNDIM

Quality Assurance in Laboratory Testing for IEM

MAY 2026

NEWSLETTER

TABLE OF CONTENTS

MESSAGE FROM THE CHAIR

Dear Colleagues,

I am pleased to welcome you to this edition of the ERNDIM newsletter, where we provide an update on current activities and developments across the organisation.

In 2026, ERNDIM experienced moderate growth in both participation and scope of activities. This year we are running 18 EQA schemes, with a total of 2,295 scheme registrations from 428 participating laboratories, compared with 417 participants in 2025. This growth is largely driven by the successful introduction of new schemes in recent years, including the Amino Acid Interpretation (AAI) scheme launched in 2023 and the Lipids in Serum (LIS) scheme, which has progressed to full scheme status this year. Work toward ERNDIM being ISO accredited is actively ongoing. An accreditation subcommittee has been formed, and a UKAS application is drafted.

ERNDIM will once again host a satellite meeting at the SSIEM Symposium, which will take place this year in Helsinki. In addition to our Diagnostic Proficiency Testing (DPT) participant workshops, the ERNDIM session will include several presentations focusing on peroxisomal disorders. Further circulated shortly. Please see the news and events page of the ERNDIM website for updates on this and other meetings:
https://www.erndim.org/news-events/

In October 2025, we held the ERNDIM Workshop in Madrid. As in previous years, we have continued the practice of organising participant meetings when the International ICIEM Congress is held outside Europe. The Madrid meeting was a great success, and we encourage readers to see the dedicated article on page 3 for further details.

Alongside in-person meetings, ERNDIM continues to offer a programme of online workshops, hosted by our Scientific Advisors and focused on technical aspects of individual schemes, primarily aimed at laboratory professionals. In March, we hosted the first ACDB participant workshop, led by Cristiano Rizzo, Erin Emmett and Joachim Janda, which focused on clinical samples from the 2024 and 2025 ACDB schemes. In April, as scientific advisor for the schemes, I also held a combined SAS and SAU technical workshop, which was well attended by participants. Slides from both workshops are available to download on our website: https://www.erndim.org/meetings-reports-cat/meetings/

I would like to take this opportunity to wish Cas Weykamp all the very best for his retirement. Cas founded MCA Laboratories Laboratories and has worked closely with ERNDIM since its inception in 1994, playing a key role in the early development of our quantitative schemes. He has been instrumental in the development of the ERNDIM results website and data evaluation processes, as well as in the production of quality comparison materials and educational panels that have supported laboratories worldwide.

Kate Straznikiewicz, Administration Assistant, left the ERNDIM Admin Team at the end of October 2025 to take up a new role with the SSIEM Administration Team. We extend our thanks to Kate for all of her hard work while working for ERNDIM and wish her the best in her new role.

2026 marks the 15 year anniversary of Sara Gardner joining ERNDIM as Executive Administrator and the founding of the ERNDIM Administration Office. Sara's appointment allowed centralisation of many administrative processes and increased support for the Scientific Advisors and Scheme Organisers. Sara has been essential to the implementation of many changes during this time, allowing ERNDIM to expand the scope and quality of the services that we provide. We would like to congratulate Sara on this milestone and thank her for her continued hard work and dedication.

Finally, I would like to emphasise that high-quality EQA schemes are only possible through the dedicated contributions of experts from across our field. On behalf of the Executive Committee, I would like to thank our Scientific Advisors for their continued enthusiastic involvement in scheme organisation and development.

Best wishes,
Rafa Artuch

On behalf of the ERNDIM Executive Committee

ERNDIM SYMPOSIUM MADRID, OCTOBER 2025

ERNDIM was delighted to welcome participants to the ERNDIM Participant Workshop, held 9–10 October 2025 in Madrid, Spain. The meeting provided an important opportunity for the international metabolic diagnostics community to meet in person and share expertise across laboratory and clinical practice.

The workshop was attended by 101 participants from 26 countries, including laboratory delegates, invited speakers and guests. The programme covered a wide range of topics relevant to the diagnosis of inborn errors of metabolism, with sessions on lysosomal metabolism, novel lipid biomarkers, rare organic acidurias, and challenging cases.

These were complemented by oral abstract sessions and interactive workshops focusing on organic acids, amino acids and lysosomal enzymes, alongside live Q&A discussions.

Feedback from the post-meeting survey was very positive. Respondents strongly agreed that the programme was relevant to their professional activity, well organised, and met their educational goals. The majority rated the overall quality, administration and value of the workshop as excellent or good. Participants particularly valued the practical focus of the sessions, the interactive workshops, and opportunities for discussion and networking with colleagues and ERNDIM representatives. Many attendees reported that the workshop would have a direct impact on their future practice, including improved interpretation skills, increased awareness of new biomarkers and diagnostic approaches, and plans to implement or refine laboratory methods.

ERNDIM would like to thank all speakers, chairs and participants for their active contribution to the workshop, and for the valuable feedback provided. Insights from the survey will help inform the planning of future educational events.

Copies of the workshop presentations, including oral and poster abstracts, are available on the ERNDIM website: https://www.erndim.org/meetings-reports/erndim-workshop-madrid-2025/

APPEAL FOR DONATED SAMPLES

ERNDIM Qualitative schemes are dependent on the availability of real patient samples. Participants are encouraged to contact ERNDIM if they are able to donate samples which may be suitable for any of the Qualitative schemes run by ERNDIM. If the sample is suitable and is used in a scheme, the donating laboratory will receive a 20% discount on their participation in the scheme, during the scheme year following use of the donated sample.

Sample requirement details and a consent form can be found on the EQA schemes tab of the ERNDIM website under “Sample Donations”. 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).

ERNDIM TRAINING SUPPORT GRANT: STRENGTHENING METABOLIC DIAGNOSTICS IN THE PHILIPPINES

ERNDIM recently awarded a Training Support Grant to Dr. Dahlia C. Apodaca, Head of the Biochemical Genetics Laboratory at the University of the Philippines–Manila. This grant enabled her to undertake short-term, hands-on training at Erasmus University Medical Center and Maastricht University Medical Center, focusing on advanced analytical techniques essential for diagnosing inborn errors of metabolism (IEM).

During her visit, Dr. Apodaca received practical and theoretical training in MS/MS-based GAG quantification, purine and pyrimidine (PUPY) analysis, method validation, and a range of biochemical assays including targeted metabolomics, oligosaccharide screening, and organic acid analysis. The experience also provided valuable insight into best laboratory practices, quality management, and approaches used in world-leading metabolic centres.

These new skills directly support the laboratory's goal of expanding diagnostic capability for both common and ultra-rare metabolic disorders in the Philippines, and will contribute to improved services for patients with IEM.

The visit additionally opened opportunities for future collaboration with Dutch experts in metabolic diagnostics and research.

Beyond the scientific programme, Dr. Apodaca highlighted the cultural value of the visit, from experiencing The Netherlands' strong commitment to environmental sustainability to enjoying its rich artistic heritage, including a memorable trip to the Rijksmuseum.

Dr. Apodaca's full reflection on the training, covering scientific outcomes, collaborations, and broader experiences can be read in the full report available on our website.

Interested in applying?

ERNDIM Training Support Grants are available, and we encourage colleagues seeking specialist training to review the eligibility criteria and application details on our website.

International Metabolomics Workshop

On January 18th and 19th 2026, the first international workshop on the use of metabolomics to diagnose inherited metabolic diseases took place in Barcelona. This workshop was organized by Judith Jans (Utrecht), Judit Garcia-Villoria (Barcelona), Cristiano Rizzo (Rome), and George Ruijter (Rotterdam), with support from ERNDIM.

Twenty participants were invited from labs that already use metabolomics in the clinic or are developing it. It was incredibly helpful and enjoyable to exchange ideas about technical aspects, sample storage, quality control, data interpretation, European regulations, and more. The workshop was also the first step to establish an international network to share experiences and knowledge to improve this new form of diagnostics.

Scheme Spotlight: Diagnostic Proficiency Testing (DPT)

The Diagnostic Proficiency Testing (DPT) scheme is one of ERNDIM's longest-running External Quality Assessment (EQA) programmes, designed to evaluate a laboratory's ability to diagnose Inherited Metabolic Diseases (IMDs) using real-world clinical scenarios.

Unlike many EQA schemes that focus on individual analytical results, DPT takes a holistic approach to assessing diagnostic performance. Each participating laboratory receives a urine sample alongside brief clinical information, mirroring the complexity and ambiguity of routine IMD investigations. From there, laboratories must select and perform the most appropriate biochemical tests, interpret the analytical findings, provide a diagnostic conclusion and recommend any additional tests needed for confirmation. This mirrors true clinical practice and offers participants a unique opportunity to evaluate and strengthen their full diagnostic process.

Recent analysis has further underlined the value of the scheme[1]. A 2022 study by Mathis et al. examining several years of DPT performance data demonstrated that ERNDIM's diagnostic proficiency schemes contribute meaningfully to improvements in diagnostic accuracy and consistency across laboratories. The authors reported that when similar disorders were re-distributed in subsequent years, laboratory performance improved in the majority of cases, highlighting DPT's role in supporting continual learning, methodological refinement, and greater clinical confidence when interpreting complex IMD presentations.

Participation in DPT requires laboratories to have access to a defined core panel of assays, including amino acids, organic acids, oligosaccharides, mucopolysaccharides, and purines and pyrimidines. While the use of cluster laboratories for certain tests is acceptable, participants remain responsible for the accuracy of all submitted results. This requirement helps ensure that all laboratories can reliably assess common IMDs encountered in clinical practice.

As a scheme that closely reflects real diagnostic workflows, DPT is highly valued for its educational impact. Each year, scientific advisors host workshops —either online or at the annual SSIEM Symposium—where participants can review cases, discuss challenges, and learn directly from experts and peers.

In recognition of the crucial role laboratories play in supplying high-quality EQA materials, ERNDIM also offers a 20% discount on DPT registration fees for participants who donated an accepted urine sample used in the previous scheme year. This incentive supports sustainability and fosters collaboration across the IMD diagnostic community.

As ERNDIM continues to enhance quality and consistency in metabolic diagnostics worldwide, the DPT scheme remains at the heart of our commitment to education, accuracy, and improved patient care.

[1] Mathis D, Croft J, Chrastina P, Fowler B, Vianey-Saban C, Ruijter GJG. The role of ERNDIM diagnostic proficiency schemes in improving the quality of diagnostic testing for inherited metabolic diseases. J Inherit Metab Dis. 2022; 45(5): 926-936. doi:10.1002/jimd.12523

2026 SSIEM Academy

ERNDIM is pleased to highlight an upcoming training opportunity from SSIEM. The 2026 SSIEM Academy Clinical and Laboratory Scientist Course will take place in Amsterdam, The Netherlands, from 9–11 November 2026, and marks the first course in a newly revised three-year curriculum cycle with an updated format.

The revised programme is designed to promote close interaction between clinicians and laboratory scientists, combining joint lectures, roundtable discussions, workshops, and hands-on data interpretation sessions. Core topics for the 2026 Academy will include amino acid disorders, urea cycle defects, organic acidemias, disorders of cobalamin metabolism, and neurotransmitter disorders.

Course applications opened on Monday 23 March 2026 and will close at 23:59 GMT on Monday 18 May 2026. Places are limited and may be subject to a selection process. Successful applicants will be invited to submit a short case abstract, with selected trainee cases presented during the Academy.

The registration fee includes accommodation, course attendance, and lunches, with an optional group dinner also available. Participants should note that travel costs are not included.

Further details on the programme can be found via the Training section of the SSIEM website. To apply, please visit the applications page of the SSIEM website, and for any other queries, please contact the SSIEM Admin Team at admin@ssiem.org.

Update on ERNDIM Quality Comparison Materials

Quality Comparison Materials have been developed by ERNDIM in collaboration with MCA Laboratories and are available via the ERNDIMQA website as an additional service for users of ERNDIM EQA schemes. The range of analytes and concentrations continues to be defined in close collaboration with the relevant scientific advisors, and may be expanded in response to participant demand.

Following the implementation of the EU In Vitro Diagnostic Medical Devices Regulation (IVDR), the regulatory requirements for Internal Control materials have become significantly stricter, including a new obligation for CE marking. Obtaining CE certification would lead to substantial cost increases, making continued provision of these materials financially unsustainable for many laboratories.

To address this, ERNDIM has decided to replace its Internal Control materials with Quality Comparison Materials. While the composition of the materials remains unchanged, their intended purpose has been modified, meaning they no longer fall under IVDR CE-marking requirements. This provides a practical and cost-effective solution, while maintaining the quality and consistency laboratories rely on.

In addition, the existing data management platform remains available for most materials through the Quality Comparison System (QCS).

Laboratories are encouraged to voluntarily submit results, as increased participation improves the reliability of the assigned values through comparison with other laboratories using the same material batch.

Please note that all orders and payments for Quality Comparison Materials are handled directly by MCA Laboratories, not by ERNDIM.

Further details are available on the ERNDIMQA website under General Information → Webshop → Quality Comparison Materials.

Quality Comparison Material	*Content of a package*	*Use QCS*
Acylcarnitines	6 vials with low concentrations and 6 vials with high concentrations	Yes
Amino Acids		Yes
Organic Acids		Yes
Pterins in Urine		Yes
Purines & Pyrimidines		Yes
Special Assays in Serum		Yes
Special Assays in Urine		Yes
Homocysteine	6 vials with a low, medium, or high concentration	No
Cystine in WBC; protein	6 vials with a low, medium, or high concentration	No
Cystine in WBC; cystine		No

Updates to the MCA Results Website

We would like to draw your attention to several recent updates to the MCA results website used for our quantitative and hybrid EQA schemes. These changes form part of our ongoing efforts to improve the platform, incorporate feedback from participants, and enhance functionality to support clearer interpretation and analysis of results.

We encourage participants to continue providing feedback on the results website, either by emailing admin@erndim.org or through the annual participant survey. We also recommend reviewing the participant survey report, where we aim to respond directly to suggestions and feedback received.

- **Excel export for cycle review data** – Participants can now export cycle review data in Excel format for easier downstream analysis. This option is available alongside the PDF export at the top of the report.
- **Directional z-scores** – Z-scores were previously reported as absolute values only. They now include a direction of effect, allowing quicker and more intuitive interpretation.
- **Units displayed in cycle review reports** – Units now appear directly in the cycle review report and are automatically appended to the analyte name.
- **Standard deviations added to cycle review** – Standard deviations are now displayed within the cycle review report, making interpretation easier without the need to open additional reports.

Analyte	Your Lab	Med All Labs	n	ZScore
2-Aminobutyric acid (µmol/L)	157	121	187	3
Alanine (µmol/L)	223	207	266	0.9
Alloisoleucine (µmol/L)	41.0	39.9	206	0.2
Arginine (µmol/L)	540	549	268	-0.3
Argininosuccinic acid (µmol/L)	5.00	5.00	147	0
Asparagine (µmol/L)	118	112	250	0.2
Aspartic Acid (µmol/L)	61.0	51.7	253	1.2
Citrulline (µmol/L)	1088	1182	265	-0.7

Participant Survey Feedback Highlights Progress and Key Challenges for ERNDIM

ERNDIM will shortly publish the results of its latest participant survey covering the 2025 scheme year, providing valuable insight into participant experiences alongside a detailed summary of External Quality Assessment (EQA) scheme performance. The findings demonstrate continued confidence in ERNDIM's services and a perception of improving performance, while also identifying specific aspects that would benefit from further attention.

The participant survey was distributed on 14 January 2026 to 828 contacts across 415 centres and closed on 11 February 2026. Responses were requested in relation to the 2025 scheme year. In total, 138 individuals from 133 centres in 41 countries participated. This corresponds to a 32% response rate by centre, representing a slight decrease of 2% compared with 2024. Despite the modest decline in engagement, the survey captured a broad international snapshot of participant opinion.

Overall sentiment towards ERNDIM's performance remains positive. A combined 67.7% of respondents reported that ERNDIM is improving, with 55.1% stating that performance is "getting better" (up from 51.7% in 2024) and 12.6% reporting it is "getting much better." Although the latter figure is lower than in the previous year (20%), the results continue to reflect a generally upward trend in perceived performance.

Satisfaction with service quality also remains high. 95.3% of respondents rated ERNDIM's services as either "excellent" or "good." While slightly lower than the 98.6% reported in 2024, this still represents a strong endorsement. Confidence in ERNDIM's ability to deliver services has increased further, with 96.9% of respondents indicating "complete" or "a lot" of confidence, compared with 95.8% in 2024.

Within the participant survey, ERNDIM also analysed feedback on individual EQA schemes. The overall score across all aspects of all schemes was 1.7, marginally worse than the 1.6 recorded in 2024, indicating a small overall decline in perceived performance.

Several aspects were rated particularly highly. The combination of sample frequency for the PPU, UMPS, and LEFB schemes received the best score of 1.4, while usefulness of the annual report consistently scored well across multiple schemes. Notably, this aspect scored 1.4 for DPT and 1.5 for ACS, ACDB QLOU, UMPS, PPU, and LEFB, reinforcing the value participants place on clear and informative reporting.

However, the analysis also highlighted recurring challenges. The "Sample volume" score for the CDG scheme was again the poorest-performing aspect, with a score of 3, worsening from 2.5 in 2024. Other lower-scoring areas included sample volume for the LEFB and SADB schemes (both scoring 2), and value for money for NCSF, SADB, CDG, and PTU, each scoring 1.9. In addition, website display for the AAI scheme was identified as a key issue, also scoring 1.9, making it the lowest overall score for this aspect.

A comparison of scheme types showed that, on average, qualitative schemes using the CSCQ website scored slightly worse (average 1.8) than quantitative and hybrid schemes using the MCA results website (average 1.7). Despite this general trend, AAI stood out as receiving the poorest overall score specifically for website display.

Taken together, the survey results indicate that while ERNDIM continues to enjoy high levels of trust and satisfaction among participants, there remain clear opportunities for improvement, particularly around sample volume, perceived value for money, and digital presentation. These insights will help guide future refinements to ERNDIM's EQA schemes and services, supporting its ongoing commitment to quality, transparency, and participant engagement.

Calls to the Community

We invite members of the community to review the following requests from ERNDIM participants. If you are able to offer advice, share resources, or lend your expertise, please reach out to the administration office at **admin@erndim.org**. Your input and collaboration are greatly appreciated.

1. Daniel Herrera (Scientific Advisor of CWBC) is **planning a case series publication on ocular cystinosis.** Any laboratory involved with at least one case of ocular cystinosis diagnosed biochemically and confirmed by genetic analysis, and wants to be involved in this publication, please contact the admin office.

2. A CDG participant in has noted that **their usual supplier of the MPSIIID substrate is no longer on the market.** Biosynth supplies the Moscerdam equivalent substrate; however, this is prohibitively expensive. Does anyone know of an alternative supplier?

3. A CDG participant is experiencing difficulties sourcing a **rabbit anti-human transferrin antibody** (Cedarlane labs) due to import issues with their usual supplier, and would welcome recommendations on alternative antibodies and suppliers for this assay.