



2022 Participant Survey Report: *[2021 scheme year]*

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1. Introduction

- Participants (808 contacts from 404 centres) were sent the link to the ERNDIM Participant Survey on the Survey Monkey website (www.surveymonkey.com) on 10th January 2022. We asked participants to answer questions relating to the 2021 EQA schemes. The closing date for the survey was 7th February 2022.

2. Summary

- Thank you to everyone who took the time to complete this survey. This report is a summary of all the responses we received. The results from the survey will help us to continue to improve the quality and efficiency of the ERNDIM EQA schemes.
- 32.9% of the laboratories that participated in the 2021 schemes responded to the survey, with the response rate for each of the schemes being between 27.8% - 54.0%.
- The survey has again highlighted areas where we need to improve, such as low sample volume for some schemes, value for money and billing arrangements.
- It is gratifying to see that 95% of respondents rate the quality of products and services we provide as 'excellent' or 'good' and that 69% of respondents believe that the quality of service we offer is getting better. We will continue to make further improvements to our services as we work towards applying for accreditation.
- The issue of sample volume is more difficult. The schemes that use real clinical samples as the EQA materials are dependent on the Scientific Advisors sourcing suitable clinical samples of sufficient volume either by direct contact with clinicians or via donations from participating laboratories. However, we are investigating alternative routes for sample donation. Information on the types of samples that would be useful to ERNDIM can be found on the website (www.erndim.org) under EQA schemes\sample donations. Discounts on scheme fees are also available for some schemes if a donated sample is used as an EQA material. If you would be interested in donating a sample, please contact admin@erndim.org for more information.
- For manufactured samples (Quantitative schemes) larger sample volumes are possible, however this would incur additional costs and as such ERNDIM aims to provide sufficient sample volume for most participants while minimising costs. For most schemes, it is possible for participants requiring a larger sample volume to purchase additional sets of samples.
- We are especially pleased that so many of you took the time to complete the survey and to send comments on the schemes. We hope you find the summary where we answer some of your comments (see page 10) and we would welcome any other comments or suggestions for improvements.

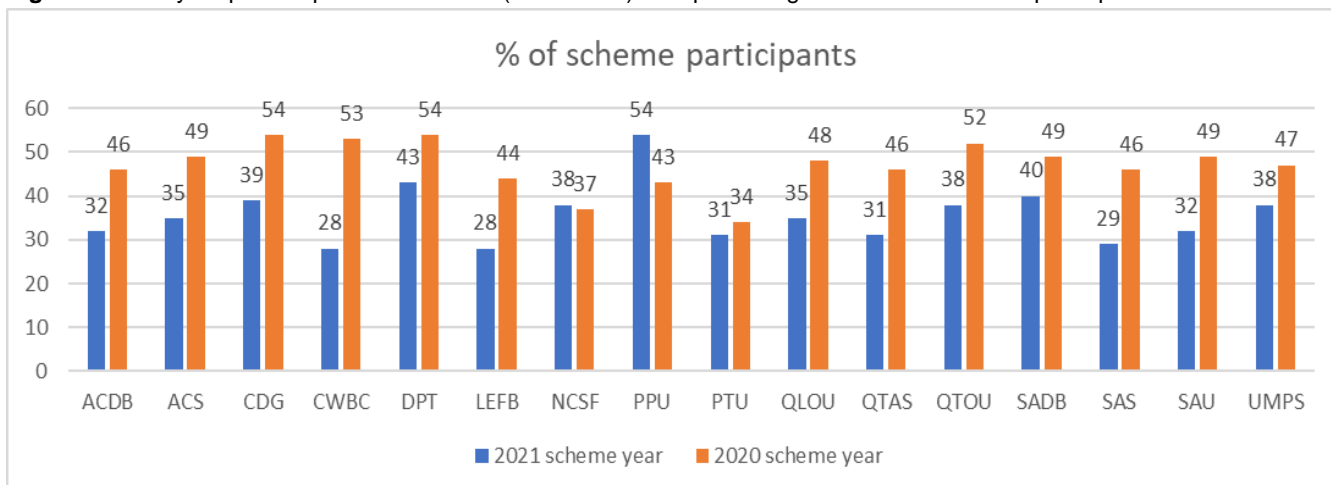
3. Survey Responses

- 137 individuals from 133 centres in 39 countries responded to the survey. The response rate by centre was 33% (compared to 46% in the last survey).

3.1. Please rate the following aspects for each of the ERNDIM quality assurance schemes that you subscribe to (Q.1 & 2)

- The response rate for each EQA scheme is shown in Figure 1 and Table 2. For the individual schemes the highest response rate was for Purines & Pyrimidines in urine (54.0% of 2021 scheme participants) and the lowest was for Cystine in WBC (27.8% of 2021 scheme participants).
- The response rate was lower for all schemes except Purines and Pyrimidines in urine and Neurotransmitters in CSF compared to 2021 with the biggest decrease being seen for Cystine in WBC (27.8% in 2021 compared to 52.6% in 2020).

Figure 1. Survey responses per EQA scheme (Question 1) as a percentage of the EQA scheme participants



Key	EQA Scheme	Code	EQA Scheme	Code
	Acylcarnitines in DBS	ACDB	Pterins in urine	PTU
	Acylcarnitines in serum	ACS	Qualitative organic acids (urine)	QLOU
	Congenital disorders of glycosylation	CDG	Quantitative amino acids (serum)	QTAS
	Cystine in white blood cells	CWBC	Quantitative organic acids (urine)	QTOU
	Diagnostic Proficiency Testing (urine)	DPT	Special assays - DBS	SADB
	Lysosomal storage enzymes (fibroblasts)	LEFB	Special assays - serum	SAS
	Neurotransmitters in CSF	NCSF	Special assays - urine	SAU
	Purines & pyrimidines (urine)	PPU	Urine Mucopolysaccharides	UMPS

- Participants were asked to rate the following aspects of each scheme:
 - Frequency of samples
 - Appropriateness of analyte concentration
 - Website display
 - Value for money
 - Sample volume
 - Adequacy of the report
 - Usefulness of the annual report
 - Billing arrangements
- Each of the aspects of individual EQA schemes was rated according to the following scoring system:
 - 1 = Excellent
 - 2 = Good
 - 3 = Poor
 - 4 = Very poor
- The average scores per scheme since 2001 are shown in Table 1 and Figure 2 and scores ≤ 1.5 are highlighted in blue and scores ≥ 2.0 are highlighted in red.
- The overall score for all aspects of all schemes was 1.7, which is the same as for the 2020 scheme year. Ten of the EQA schemes had the same score as last year, one scheme had a worse score than last year (PPU) and 5 schemes had better scores (CDG, LEFB, NCSF, QTOU and UMPS).
- The best scoring schemes were DPT, NCSF, PTU and UMPS scoring 1.6. The CDG scheme scored worst with a score of 1.8, however this is an improvement from 1.9 for the 2020 CDG scheme.
- The score for the most individual aspects remained unchanged when compared to the 2020 scheme year, see Table 2. The score for 'Billing arrangements' improved from 1.8 for the 2020 scheme year to 1.7 for 2021. The score for 'Website display' worsened from 1.7 for the 2020 scheme year to 1.8 for 2021.
- The worst scoring aspects were 'Sample volume', 'Website display', and 'Value for money' having an average score of 1.8. The best scoring aspects were 'Frequency of samples', 'Adequacy of the report' and 'Usefulness of the annual report' which all scored 1.6.

Table 1. Average scores per scheme (Question 1) [See Figure 1 for key to scheme codes]

EQA Scheme	Average Scores												
	2021	2020	2019	2018	2017	2016	2015	2014	2013	2012	2011	2007	2001
All schemes	1.7	1.7	1.7	1.8	1.7	1.7	1.7	1.8	1.7	1.7	1.7	1.8	1.7
ACDB	1.7	1.7	1.7	1.8	1.8	1.8	1.9	1.9	2.0	1.9	1.9	2.0	2.0
ACS	1.7	1.7	1.7	1.7	1.6	-	-	-	-	-	-	-	-
CDG	1.8	1.9	1.9	1.9	1.8	1.9	1.9	2.0	2.0	1.9	1.8	1.9	-
CWBC	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.8	1.8	1.6	1.7	1.6	1.4
DPT	1.6	1.6	1.7	1.8	1.6	1.7	1.7	1.7	1.7	1.7	1.8	1.8	1.7
LEFB	1.7	1.8	1.9	1.8	1.7	1.8	1.9	1.9	2.0	1.9	2.0	2.1	-
NCSF	1.6	1.9	1.8	1.8	1.9	1.7	-	-	-	-	-	-	-
PPU	1.7	1.6	1.7	1.7	1.7	1.7	1.8	1.8	1.7	1.7	1.7	1.9	1.6
PTU	1.6	1.6	1.5	1.8	1.9	-	-	-	-	-	-	-	-
QLOU	1.7	1.7	1.6	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.6
QTAS	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.8	1.7
QTOU	1.7	1.8	1.7	1.8	1.7	1.7	1.7	1.8	1.7	1.7	1.7	1.9	1.7
SADB	1.7	1.7	1.8	-	-	-	-	-	-	-	-	-	-
SAS	1.7	1.7	1.7	1.7	1.7	1.8	1.8	1.7	1.7	1.7	1.7	1.8	1.7
SAU	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.8
UMPS	1.6	1.7	1.7	1.8	1.7	1.8	1.7	1.8	1.8	1.8	1.8	-	-

- There was a total of 3 scores of 2.0 or more in this survey: CDG ('Sample volume' = 2.5 and 'Website display' = 2.1) and NCSF ('Sample volume' = 2.1).
- The 'Sample volume' score for CDG was again the worst score in the survey (2.5) with a slightly improved score than for 2020 and 2019 scheme years (2.6 for 2020 and 2.4 for 2019).
- The best scores of the whole survey (1.4) were for 'Website display' for PTU and both 'Adequacy of the report' and 'Usefulness of the Annual Report' for DPT.

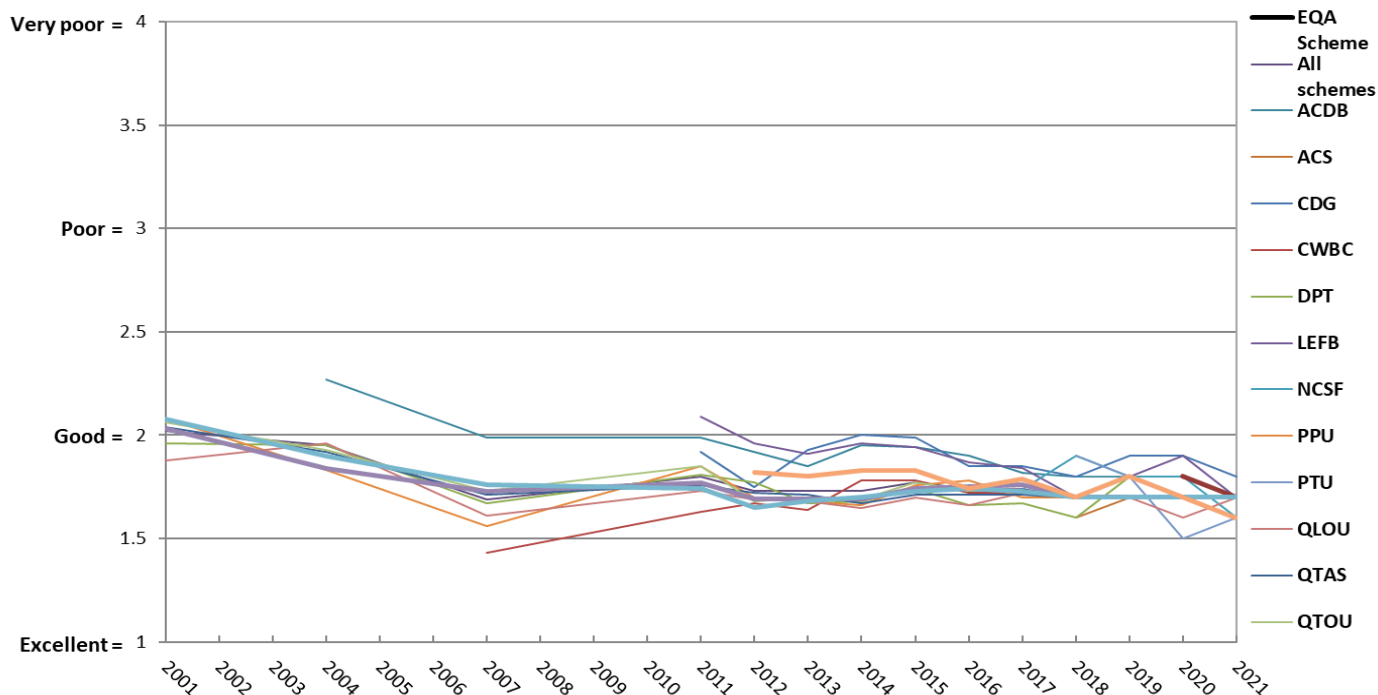


Figure 2. Average score per EQA scheme (Question 1) [See Figure 1 for key to scheme codes]

Table 2: Average scores per aspect of each scheme (Question 1) [See Figure 1 for key to scheme codes]

Scheme Aspects	Frequency of samples	Sample volume	Appropriateness of analyte concentration	Adequacy of the report	Website display	Usefulness of the annual report	Value for money	Billing arrangements	Average per scheme	No. of responses (% of scheme participants)
EQA Schemes										
ACDB	1.7	1.8	-	1.6	1.9	1.6	1.8	1.7	1.7	42 (31.8%)
ACS	1.5	1.5	1.7	1.7	1.8	1.6	1.8	1.7	1.7	44 (34.6%)
CDG	1.5	2.5	-	1.7	2.1	1.5	1.7	1.7	1.8	26 (38.8%)
CWBC	1.7	2.1	1.6	1.6	1.7	1.8	1.6	1.8	1.7	10 (27.8%)
DPT	1.5	1.8	-	1.4	1.9	1.4	1.7	1.7	1.6	44 (43.1%)
LEFB	1.6	1.7	1.6	1.7	1.8	1.6	1.8	1.8	1.7	19 (27.5%)
NCSF	1.6	1.8	1.5	1.5	1.5	1.5	1.7	1.7	1.6	14 (35.1%)
PPU	1.6	1.5	1.7	1.6	1.9	1.6	1.8	1.8	1.7	27 (54.0%)
PTU	1.5	1.8	1.9	1.6	1.4	1.5	1.7	1.6	1.6	11 (31.4%)
QLOU	1.6	1.9	-	1.5	1.8	1.5	1.8	1.7	1.7	77 (34.7%)
QTAS	1.5	1.5	1.7	1.7	1.7	1.6	1.8	1.7	1.7	82 (31.3%)
QTOU	1.5	1.5	1.8	1.8	1.8	1.6	1.9	1.7	1.7	48 (37.8%)
SADB	1.6	1.6	1.7	1.7	1.8	1.7	1.8	1.7	1.7	38 (40.4%)
SAS	1.5	1.6	1.7	1.7	1.7	1.6	1.9	1.8	1.7	66 (28.7%)
SAU	1.5	1.6	1.7	1.6	1.8	1.6	1.8	1.6	1.7	60 (32.4%)
UMPS	1.5	1.9	-	1.5	1.8	1.5	1.7	1.6	1.6	33 (37.9%)
Average for all schemes	1.6	1.8	1.7	1.6	1.8	1.6	1.8	1.7	1.7	133 (32.9%)

3.2. Analytes in Quantitative Schemes (Q5 – Q.14)

- A total of 58 individuals (42.3% of respondents) made suggestions for analytes to be added to or removed from the Quantitative schemes.
- Where possible we do try to incorporate suggestions for additional analytes but unfortunately this is not always possible. A summary of the suggestions for analytes to added or removed, with some responses from ERNDIM, is below (pages 5 to 8).

Q.5: Acylcarnitines – Serum (9 responses, 7.1% of ACS participants)

Suggested Analytes to be added

Total suggested = 18

Analytes with >1 response

C16:1OH 2

Suggested Analytes to be removed

Total suggested = 1

All Analytes suggested

Total Carnitine 2

ERNDIM Response:

- No analytes were requested by a large number of participants. At this time no further will be added to the scheme as it was previously agreed by the Scientific Advisory Board (SAB) that it is important to manage the addition of analytes carefully as new additions may affect the stability of the samples due to possible cross reactions.

Q.6: Lysosomal Enzymes (11 responses, 15.9% of LEFB participants)**Suggested Analytes to be added**

Total suggested = 10

Analytes with >1 response

Arylsulfatase A	3
Change enzymes each year	2

Suggested Analytes to be removed

Total suggested = 2

All Analytes suggested

Aspartylglucosaminidase	3
Spingomyelinase	2

ERNDIM Response:

- The 2019 LEFB scheme saw the first change to the enzymes included for several years. It is the intention of the Scientific Advisor for this scheme to review the performance and requests of participants each year and adjust the scheme to address enzymes which cause difficulty or are of interest to our participants. It is hoped that a wider selection of enzymes will be included in this scheme over several years by rotating some enzymes each year and requests from participants will be considered.

Q.7: Neurotransmitters – CSF (3 responses, 8.1% of all NCSF participants)**Suggested Analytes to be added**

Total suggested = 3

Analytes with >1 response

5 Methylene tetrahydrofolate	2
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Suggested Analytes to be removed

Total suggested = 0

All Analytes suggested**ERNDIM Response:**

- 5 Methylene tetrahydrofolate is currently not requested by sufficient participants to be considered but may be revisited in the future if larger numbers of requests are received.

Q.8: Purines & pyrimidines (7 responses, 14.0% of PPU participants)**Suggested Analytes to be added**

Total suggested = 6

Analytes with >1 response

SAICAR	3
2,8-dihydroxyadenine	2
beta-Ureidobutyric acid	2

Suggested Analytes to be removed

Total suggested = 1

All Analytes suggested

Carnitine	1
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ERNDIM Response:

- SAICAR will be included for the 2023 scheme year following requests over several years for this analyte.
- 2,8-dihydroxyadenine has been attempted by the scheme organiser and Scientific Advisor but was chemically not possible.
- Beta-Ureidobutyric acid will be included in the 2023 scheme.
- Carnitine will not be removed from the scheme due to only one request.

Q.9: Pterins – Urine (1 response, 2.9% of all PTU participants)**Suggested Analytes to be added**

Total suggested = 1

All analytes suggested

Sepiapterin	1
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Suggested Analytes to be removed

Total suggested = 0

All Analytes suggested**ERNDIM Response:**

- No analytes were requested by a large number of participants.

Q.10: Quantitative amino acids (19 responses, 7.3% of all QTAS participants)**Suggested Analytes to be added**

Total suggested = 12

Analytes with >1 response

Beta-alanine	2
Phosphoethanolamine	2

Suggested Analytes to be removed

Total suggested = 10

Analytes with >3 response

N(pros)-methylhistidine	6
N(tele)-Methylhistidine	6
Homocitrulline	5
Aspartyl glucosamine	5

ERNDIM Response:

- N(pros)-methylhistidine and N(tele)-Methylhistidine will remain in the scheme. While it is understood that these analytes have caused difficulty for some labs the Scientific Advisory Board feel it is important that this is addressed rather than removing the analytes.
- Neither analyte suggested for addition to the scheme were requested by a large number of participants. At this time no further will be added to the scheme as it was agreed by the Scientific Advisory Board that it is important to manage the addition of analytes carefully as new additions may affect the stability of the samples due to possible cross reactions.

Q.11: Quantitative organic acids (10 responses, 7.9% of all QTOU participants)**Suggested Analytes to be added**

Total suggested = 17

Analytes with >2 response

succinic acid	3
Succinylacetone	2

Suggested Analytes to be removed

Total suggested = 3

All Analytes suggested

3-OH-Glutaric acid	1
Mevalonic acid	1
Suberylglycine	1

ERNDIM Response:

- There were not enough requests for addition or removal of any other analyte to justify changes.

Q.12: Special assays – Dried Blood Spots (13 responses, 13.8% of all SADB participants)**Suggested Analytes to be added**

Total suggested = 27

Analytes with >2 response

methylcitric acid	3
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Suggested Analytes to be removed

Total suggested = 3

All Analytes suggested

allo isoleucine	1
CO	1
free carnitine	1

ERNDIM Response:

- Methylcitric acid will not be added for 2023 due to too few requests.
- Allo-isoleucine and carnitine would not be considered for removal, as these are diagnostic metabolites relevant to new-born screening second tier tests and have limited alternative EQA.

Q.13: Special assays – serum (17 responses, 7.4% of all SAS participants)**Suggested Analytes to be added**

Total suggested = 30

Analytes with >1 response

Acetoacetate	2
Alpha-galactosidase A	2
C26:0 LPC	2
NTBC (nitisone)	2
Sitosterol	2

Suggested Analytes to be removed

Total suggested = 2

All Analytes suggested

Biotinidase	1
NEFA	1

ERNDIM Response:Suggested additions

- C26:0 LPC will be included in the scheme for 2023 as the Scientific Advisor agreed this may be of interest to a larger number of participants.
- The remaining requests were not considered to be of interest at this time as there were too few requests.
- Biotinidase and NEFA are present in the matrix and as such cannot be removed.

Q.14: Special assays – urine (11 responses, 5.9% of all SAU participants)**Suggested Analytes to be added**

Total suggested = 17

Analytes with >1 response

Citrate	2
Gb3	2

Suggested Analytes to be removed

Total suggested = 1

All Analytes suggested

5-Ala	1
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ERNDIM Response:

- There were too few requests for any analyte to be added or removed from the scheme at this time.

3.3. Do you have any other remarks, comments or suggestions for any of the schemes you subscribed to? (Q.15)

- Number of individual responses = 41 (29.9% of all responses).
- These comments are summarised under 3.9 (page 10) with the comments made in response to Q.21 (see page 10).

3.4. Do you think that assessment of interpretation to the Pterin in urine scheme adds value to the scheme? (Q.3)

- 13 respondents (37.1% of participants in the PTU scheme) answered this question.
- The response options were 'It adds a lot of value' (4 respondents, 30.8%), 'It adds some value' (7 respondents, 53.8%), 'It makes no noticeable difference' (2 respondents, 15.4%), 'It detracts some value' (0 respondents) or 'It detracts a lot of value' (0 respondents).

3.5. Do you think that assessment of interpretation to the Cystine in WBC scheme adds value to the scheme? (Q.4)

- 13 respondents (36.1% of participants in the CWBC scheme) answered this question.
- The response options were 'It adds a lot of value' (4 respondents, 30.8%), 'It adds some value' (7 respondents, 53.8%), 'It makes no noticeable difference' (2 respondents, 15.4%), 'It detracts some value' (0 respondents) or 'It detracts a lot of value' (0 respondents).

3.6. Does your laboratory use any of the Internal Control Materials provided by MCA laboratories? (Q.16)

- 128/137 (93.4%) respondents answered this question

Response	Number of respondents
Yes	57 (44.5%)
No	52 (40.6%)
No, but we may use these in the future	19 (14.8%)

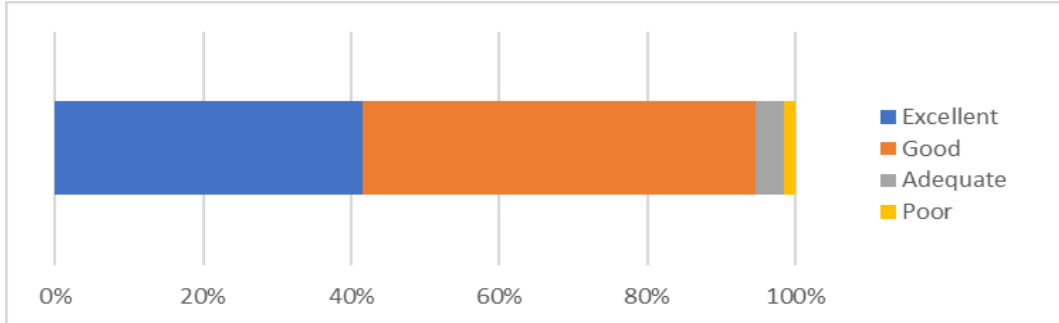
3.7. Comments on the overall performance of ERNDIM (Q.17 – 20)

- The aim of this section is to assess participants' perception of the overall performance of ERNDIM.
- In summary:
 - 95% of respondents rated the quality of services provided by ERNDIM as 'excellent' or 'good'; with 96% of respondents having 'complete' or 'a lot' of confidence that ERNDIM can deliver the service required by participants.

- 69% of respondents agreed that overall ERNDIM's performance is 'getting better' or 'getting much better'; with 95% of respondents stating that it was 'certain' or 'very likely' that they would use ERNDIM services in the future.

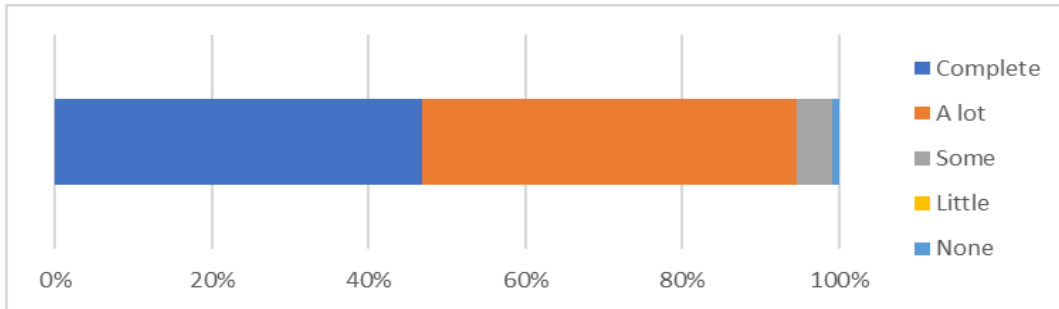
Q.17: Overall, how do you rate the quality of products and services we provide?

(130 individual responses, 94.9% of all responses for this section)



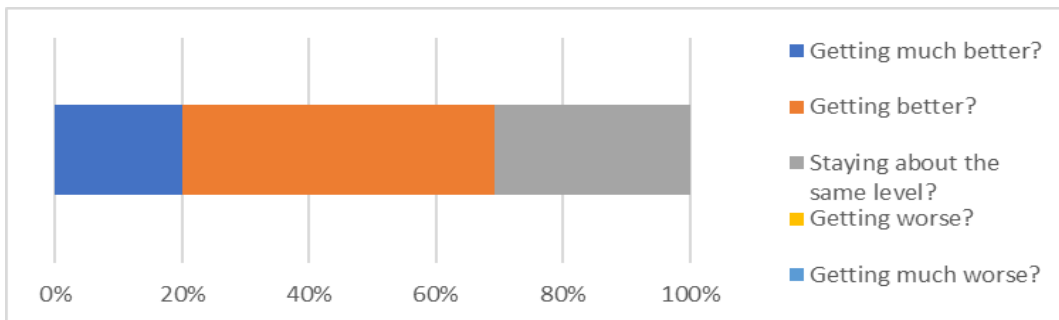
Q.18: What level of confidence do you have in us to deliver the products and services that you require?

(130 individual responses, 94.9% of all responses for this section)



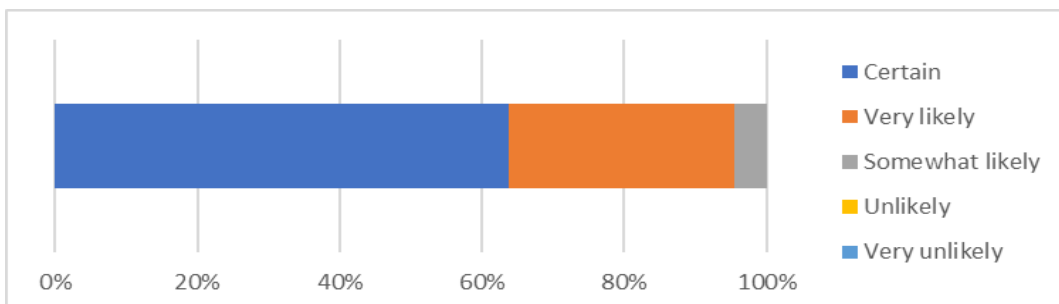
Q.19: Overall, is our performance...

(130 individual responses, 94.9% of all responses for this section)



Q.20: Based on our performance, how likely is it that you will use us in the future?

(130 individual responses, 94.9% of all responses)



3.8. Do you have any other remarks, comments or suggestions for how we could improve the services we provide? (Q.21)

- Number of individual responses = 23 (16.7% of all responses).
- These comments are summarised below with the comments made in response to Q15.

3.9. Summary of Remarks, comments or suggestions for improvements (Q.15 & Q.21)

- Total number of responses was 66 from 41 individuals (= 30% of all responses).
- There were a large number of comments and suggestions for improvement. Below is a summary of some of the most frequent comments with responses from ERNDIM.

Participant Comment

ERNDIM Response

1. Administration

- Difficulty was encountered in paying the bill.
- Periodic emails about other QC samples that ERNDIM sells would be useful.
- Qualitative Organic Acid samples were received at ambient temperature. Storage instructions were contradictory. Shipping container indicated to keep at 4°C and stated "DO NOT FREEZE". Instructions inside indicated to store the samples at -20°C.

- We are sorry to hear that any participant experienced difficulties in paying their invoice. The majority appear to do so without issue, where difficulties do arise, please contact the Administration Office as soon as possible so that we can assist.
- We will keep this in mind and consider sending more routine updates, however we endeavour to avoid overloading participants with frequent emails not relating directly to scheme participation.
- We will discuss this with the scheme organiser responsible for preparation and shipping of these samples to ensure all directions are clearer for the 2023 scheme year.

2. EQA Schemes

2.1. General

- Overall poor performance should be based on 2 consecutive years of a poor performance in the same scheme (or 2 out of 3 years) rather than 2 consecutive years of any scheme.
- Registration 2022 was not easy. We don't receive any confirmation. Maybe an automatic renewal will be better with a confirmation of the client.

- Poor performance (PP, poor performance within one scheme in a year) and Persistent PP (poor performance within one scheme for 2 out of 3 participating years) both apply only to the performance of a participant within one scheme. However we do also look at performance across schemes (Global PP and Persistent Global PP, see below) so that trends can be identified and support offered to participants that may be having wider performance issue. Performance support letters are sent to all participants identified as poor performers and are intended to offer support with an aim of improvement in performance for the benefit of patients.
- Global PP is poor performance in more than one scheme in the same participation year.
- Persistent Global PP is a poor performance in multiple schemes in 2 out of 3 years of participation. This is not necessarily PP in the same schemes each year. This is intended to bring attention to a global issue with EQA performance.
- Automatic renewals are not possible as participants need to check and confirm all contact and address details as well as confirming their EQA order.
- Registration for the 2023 scheme year will be via the new registration website, we hope that this will make the process easier for participants and welcome feedback. We apologise for difficulties with registration while our new website was under development.

2.2. Website reporting

- Why are there separate websites for the qualitative and quantitative proficiency tests? Would it be possible to have one platform where you have access to all your data?
- The functionality of the results submission websites for Qualitative and Quantitative schemes for scoring is quite distinct. At the time of creating these websites the most appropriate hosts were contracted to deliver these websites. There are no plans to merge these websites as this would require a large investment of resources. ERNDIM is currently prioritising improvement of schemes and progress towards accreditation, however, result submission website redevelopment may be considered in the future.

Participant Comment

ERNDIM Response

2.3. Acylcarnitines in DBS

- For the Acylcarnitines in DBS, the results feedback from the scheme in charge or advisor should be a little quicker just as fast as the Quantitative Serum Amino acids scheme. Otherwise, the feedback is excellent and informative.

- Scoring and preparation of interim and final reports for the qualitative schemes, including ACDB, require in depth review by the Scientific Advisor due to the Qualitative nature of interpretation in comparison to the Quantitative schemes. Development of the scoring and reporting software is underway for the ACDB scheme, as already in use for DPT and we hope that this will help to reduce this workload and ensure reports are available promptly in the future.

2.4. Acylcarnitines in serum

- In Acylcarnitines quantitative Scheme, would be great to have analyte names with abbreviation mentioned eg. Tiglylcarnitine (C5:1). This would make the data input process lucid!

- The website manager has made this change.

2.5. CDG scheme

- Faster appearance of the CDG Annual Report.

- As for the ACDB scheme we hope that development of the scoring and reporting software will make this a quicker process in 2023.

2.6. Cystine in white blood cells

- Practical issue with CWBC SNT samples: small volume, sample often in the tube cap, tube format not suitable for spinning.

- We have not previously received any concerns about this issue, if this is reported by other participants, we will review the type of tube used.

2.7. Lysosomal Enzymes in fibroblasts scheme

- Since most of the laboratories are preferring lysosomal enzyme testing on DBS due to the easy transportation from distant places, stability of enzyme and less biohazard, DBS scheme for these assays at least for 5-6 common disorders (Gaucher, Pompe, Hurler, Hunter, Fabry, Niemann Pick, GM1) should be started.

- A lysosomal enzymes in DBS pilot scheme is under consideration by ERNDIM but this requires donation of suitable samples for use as the EQA materials.

2.8. Pterins in urine

- We experienced problems in the interpretation of urine pterin profiles, with the possibility of 2 diagnosis.

- We apologise for difficulties caused by this issue. Scoring of the scheme was adjusted to ensure no participants were scored poorly as a result of the sample issue.

2.9. Qualitative Organic Acids

- If possible, to please revert to distribution of 9 urine samples for qualitative organic acids in urine, instead of 6 only.

- The change from 9 samples to 6 samples was introduced due to difficulties in collecting sufficient samples of a large enough volume to deliver 9 samples from all 3 QLOU centres each year.
- For information regarding sample donation please visit: <https://www.erndim.org/eqa-schemes/sample-donations/>

2.10. Quantitative Amino acids in serum

- Could the pdf amino acid report contain the histograms for each amino acid in addition to the summary sheet?

- The Quantitative schemes result submission website has an "analyte in detail" report which includes the histogram showing the distribution of submitted results for individual analytes within a sample. The report allows pdf printing for each analyte or for all analytes within a sample.

2.11. Quantitative Organic acids in urine

- We would like to have a wider range of concentrations for amino and organic acids.

- The Scientific Advisors for each scheme aim to select analyte concentrations which may be found in real clinical samples. However, this does also consider the practicalities of preparing these samples with a large number of analytes at varying concentrations and as such some concentrations may not always be possible.

Participant Comment

- A problem for this program is the concentration of the analytes such as the frequent unphysiological concentrations (e.g. very low TCA intermediates) which suggests the samples are not really representative of true patient samples. Moreover, does it make sense to quantify values below the URL with the same precision as around the URL or above?

2.12. Special Assays in DBS

- Blood spot special assays really needs improvement with more sample volume! Should consider the need for a separate blood spot enzyme scheme.

2.13. Special Assays in serum

- For SAS scheme, variable concentrations of NEFA (and, to a lesser extent, cholesterol) among samples would be more informative.
- Special assays in serum - more varied biotinidase concentrations, very high homocysteine concentrations (we recently identified an issue with our assay with very high homocysteine samples, ~400umol/L, which had not been picked up until we saw an affected patient sample).

2.14. Special Assays in urine

- The cystine levels in the SAU scheme seems a lot of normal or low levels. We use the assay to monitor patients with cystinuria to check the concentration of cystine is kept below 1000 umol/L to reduce stone formation so it would be more useful to us if most EQA samples for urine cystine were in the range of 700 -1300 umol/L. Many Thanks.

2.15. Urine Mucopolysaccharides

- Some samples of Mucopolysaccharides in urine hardly can be evaluated as the MPS-fractions cannot be separated adequately. Samples look like contaminated.

ERNDIM Response

- For some compounds known to be important at small concentrations, like mevalonic acid or 3-OH glutaric acid, the SA requested concentrations of 0,5,10 and 0,10,20 respectively.
TCA intermediates:
Fumaric acid: spiked 0, 20, 50, 150 (R.I. <22 for neonate and <1 for adults)
Keto-glutaric acid: 0, 100, 300, 500 (R.I. <399 for neonate and <69 for adults)

- The sample size is suitable for most participants, and providing a larger sample would require an increase in participation cost. For labs that do require larger sample size it is usually possible to purchase a second set of samples during registration. A separate lysosomal enzymes in DBS pilot scheme is under consideration by ERNDIM.

- NEFA is present in the sample matrix and is not added as a spiked analyte, so it is not possible to vary its concentration in the samples.
- A variety of concentrations are included and may vary between scheme years. For example, Homocysteine spiked concentrations have ranged from 0 to 192 umol/L.

- A wider range of concentrations including higher concentrations, has been included in the 2022 scheme.

- The majority of participants do not appear to have difficulties with these samples. If you require some advice or support, please contact admin@erndim.org and your request will be passed to the Scientific Advisor for the scheme.

3.10. Please complete your name and institute address details (Q.22-23)

- Number of individual responses = 80 (58% of all responses).