ERNDIM

QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

2020 Participant Survey Report:

[2019 scheme year]

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1. Introduction

- Participants (818 contacts from 409 centres) were sent the link to the ERNDIM Participant Survey on the Survey Monkey website (www.surveymonkey.com) on 17th January 2020. We asked participants to answer questions relating to the 2019 EQA schemes. The closing date for the survey was 17th February 2020.

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.
- 50.4% of the laboratories that participated in the 2019 schemes responded to the survey, with the response rate for each of the schemes being between 41.2% - 61.1%.
- The survey has again highlighted areas where we need to improve, such as low sample volume for some of the qualitative schemes. Some participants are also unhappy with the analyte concentrations in some schemes and specific comments from ERNDIM for the relevant schemes can be found in the summary of 'Remarks, comments or suggestions for improvements' on page 10.
- However, it is gratifying to see that 96% 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 the service that we offer 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.
- 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, interesting (see page 10) and we would welcome any other comments or suggestions for improvements.

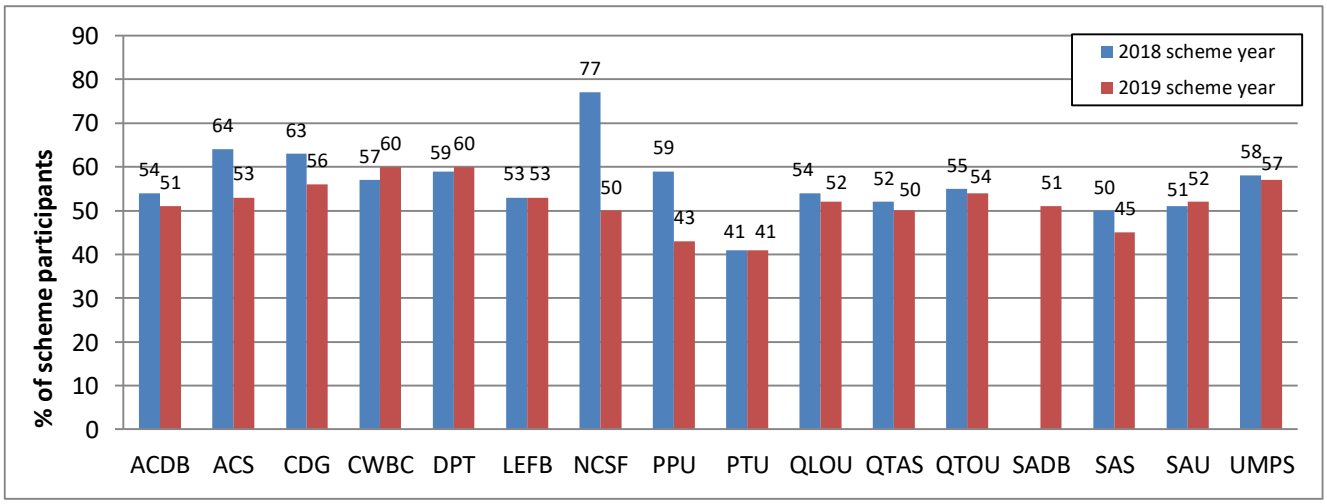
3. Survey Responses

- 217 contacts from 206 centres in 51 countries responded to the survey. The response rate by centre was 50% (compared to 53% 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)

- 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 Cystine in White Blood Cells (61.1% of 2019 scheme participants) and the lowest was for Pterins in urine (41.2% of 2019 scheme participants).
- The response rate was higher for 7 schemes than in 2019, and lower for 9 schemes compared to 2019 with the biggest decrease being seen for Purines and Pyrimidines (42.9% in 2020 compared to 59.3% in 2019). The highest response rate was for Urine Mucopolysaccharides (57.3% of scheme participants) and the lowest was for Purines and Pyrimidines (42.9% of scheme participants).

Figure 1. Survey responses per EQA scheme (Question 1) as a percentage of the EQA scheme participants *[n.b. 2019 was the first year that the SADB scheme ran as a full EQA scheme]*



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 slightly better than in 2019 (1.8). Eight of the EQA schemes had the same score as last year, one scheme had a worse score than last year (Lysosomal Enzymes in fibroblasts, 1.9 compared to 1.8 in 2019) and 6 schemes had better scores (ACDB, DPT, PTU, QLOU, QTOU and UMPS).
- The best scoring scheme was PTU which scored 1.5. The worst scoring schemes were the CDG and Lysosomal Enzymes in fibroblasts schemes which both scored 1.9.
- The scores for each scheme in each of the individual aspects are given in Table 2. The score for 6 out of the 8 of the individual aspects improved since last year, while the scores for both 'Value for money' and 'Billing arrangements' remained unchanged.
- The worst scoring aspects were 'Value for money' and 'Billing arrangements' with both 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]*

	Average Scores												
EQA Scheme	**2020**	**2019**	**2018**	**2017**	**2016**	**2015**	**2014**	**2013**	**2012**	**2011**	**2007**	**2004**	**2001**
All schemes	**1.7**	**1.8**	**1.7**	**1.7**	**1.7**	**1.8**	**1.7**	**1.7**	**1.7**	**1.8**	**1.7**	**2.0**	**2.0**
ACDB	1.7	1.8	1.8	1.8	1.9	1.9	2.0	1.9	1.9	2.0	2.0	2.3	-
ACS	1.7	1.7	1.6	-	-	-	-	-	-	-	-	-	-
CDG	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	1.8	1.8	1.6	1.7	1.6	1.4	-	-
DPT	1.7	1.8	1.6	1.7	1.7	1.7	1.7	1.7	1.8	1.8	1.7	2.0	2.0
LEFB	1.9	1.8	1.7	1.8	1.9	1.9	2.0	1.9	2.0	2.1	-	-	-
NCSF	1.8	1.8	1.9	1.7	-	-	-	-	-	-	-	-	-
PPU	1.7	1.7	1.7	1.7	1.8	1.8	1.7	1.7	1.7	1.9	1.6	1.8	2.1
PTU	1.5	1.8	1.9	-	-	-	-	-	-	-	-	-	-
QLOU	1.6	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.6	2.0	1.9
QTAS	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.8	1.7	1.9	2.0
QTOU	1.7	1.8	1.7	1.7	1.7	1.8	1.7	1.7	1.7	1.9	1.7	1.9	2.1
SADB	1.8	-	-	-	-	-	-	-	-	-	-	-	-
SAS	1.7	1.7	1.7	1.8	1.8	1.7	1.7	1.7	1.7	1.8	1.7	1.8	2.0
SAU	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.7	1.8	1.9	2.1
UMPS	1.7	1.8	1.7	1.8	1.7	1.8	1.8	1.8	1.8	-	-	-	-

- There were a total of 7 scores of 2.0 or more in this survey: CDG ('Sample volume' = 2.4), CDG ('Website display'=2.0), CWBC ('Sample volume' = 2.0), CWBC ('Billing arrangements'=2.0), LEFB ('Sample volume' = 2.0), LEFB ('Website display'=2.0) and LEFB ('Billing arrangements'=2.0).
- The 'Sample volume' score for CDG was again the worst score in the survey scoring the same as in 2019 (2.4 in 2020 and 2019).
- The best scores of the whole survey (1.3) were for 'Adequacy of the report' and 'Website display', both for Pterins in urine.

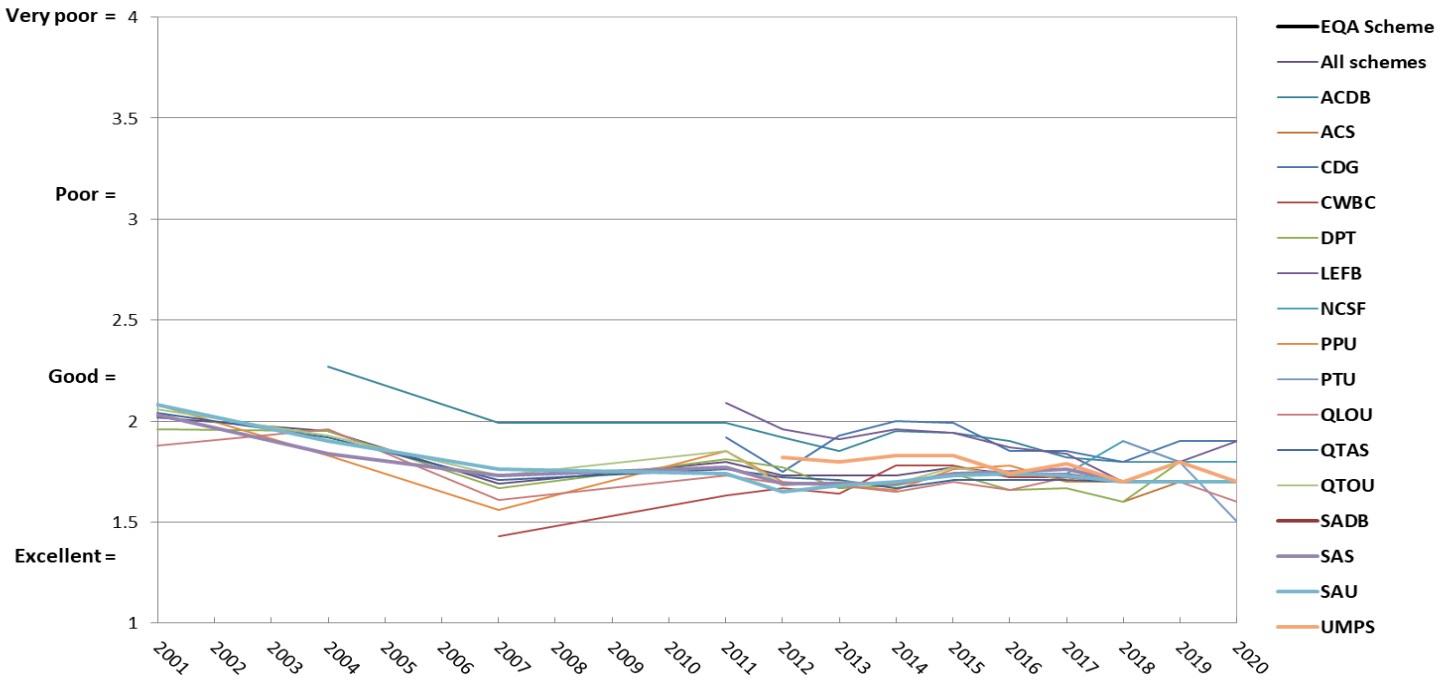


Figure 2. Average score per EQA scheme (Question 1) *[See Figure 1 for key to scheme codes]*

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

Scheme Aspects / EQA Schemes	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)
ACDB	1.7	1.7	-	1.6	1.7	1.5	1.8	1.8	1.7	71 (51.5%)
ACS	1.5	1.6	1.6	1.6	1.7	1.6	1.7	1.8	1.7	66 (54.1%)
CDG	1.6	2.4	-	1.7	2.0	1.7	1.8	1.8	1.9	38 (56.7%)
CWBC	1.6	2.0	1.7	1.6	1.6	1.6	1.8	2.0	1.7	22 (61.1%)
DPT	1.6	1.8	-	1.4	1.8	1.4	1.7	1.8	1.7	66 (60.2%)
LEFB	1.8	2.0	1.7	1.9	1.8	1.8	2.0	2.0	1.9	40 (54.8%)
NCSF	1.7	1.9	1.8	1.8	1.6	1.7	1.8	1.8	1.8	17 (50.0%)
PPU	1.6	1.5	1.7	1.7	1.7	1.7	1.8	1.8	1.7	24 (42.9%)
PTU	1.4	1.6	1.5	1.3	1.3	1.4	1.9	1.7	1.5	14 (41.2%)
QLOU	1.6	1.8	-	1.5	1.7	1.5	1.7	1.7	1.6	122 (53.0%)
QTAS	1.5	1.5	1.6	1.6	1.6	1.6	1.8	1.8	1.7	137 (50.9%)
QTOU	1.6	1.6	1.7	1.7	1.7	1.7	1.8	1.7	1.7	71 (55.5%)
SADB	1.7	1.7	1.7	1.7	1.8	1.8	1.9	1.8	1.8	44 (51.2%)
SAS	1.6	1.6	1.7	1.6	1.6	1.7	1.8	1.9	1.7	110 (46.6%)
SAU	1.6	1.6	1.7	1.6	1.6	1.6	1.8	1.9	1.7	94 (53.1%)
UMPS	1.6	1.8	-	1.6	1.7	1.5	1.8	1.7	1.7	57 (57.3%)
Average for all schemes	**1.6**	**1.8**	**1.7**	**1.6**	**1.7**	**1.6**	**1.8**	**1.8**	**1.7**	**217 (50.4%)**

3.2. Analytes in Quantitative Schemes (Q3 – Q.12)

- A total of 89 individuals (41%) 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.3: Acylcarnitines – Serum (11 responses, 5.1% of all respondents)

Suggested Analytes to be added

Total suggested = 21

Analytes with >1 response

C10:1	2
C12	3

Suggested Analytes to be removed

Total suggested = 4

All Analytes suggested

C10	1
C4-DC	1
C4-OH	1
Total carnitine	1

ERNDIM Response:

- Neither analytes were requested by a large number of participants. At this time neither will be added to the scheme as it was agreed by the ERNDIM 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.4: Lysosomal Enzymes (20 responses, 9.2% of all respondents)

Suggested Analytes to be added		Suggested Analytes to be removed	
Total suggested = 22		Total suggested = 3	
Analytes with >1 response		**All Analytes suggested**	
Alfa iduronidase	4	Palmitoyl protein thioesterase	3
Alpha-fucosidase	2	Lysosomal acid lipase (LAL/ acid esterase)	5
Alpha-mannosidase	2	Tripeptidyl peptidase I	3
Arylsulfatase B	5		
Beta-mannosidase	2		
Hexosaminidase	8		
Mannosidase	2		
MPS enzymes	3		
Sphingomyelinase	3		

ERNDIM Response:

- The 2019 LEFB scheme has seen 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.

Q.5: Neurotransmitters – CSF (4 responses, 1.8% of all respondents)

Suggested Analytes to be added		Suggested Analytes to be removed	
Total suggested = 4		Total suggested = 1	
Analytes with >1 response		**All Analytes suggested**	
MHPG	2	HVA:5-HIAA ratio	1

ERNDIM Response:

- MHPG is currently not requested by sufficient participants to be considered but may be revisited in the future if larger numbers of requests are received.
- The HVA:5-HIAA ratio will remain as the removal request is from one participant only.

Q.6: Purines & pyrimidines (6 responses, 2.8% of all respondents)

Suggested Analytes to be added		Suggested Analytes to be removed	
Total suggested = 5		Total suggested = 1	
Analytes with >1 response		**All Analytes suggested**	
SAICAR	3	2,8-dihydroxyadenine	1
Dihydroxyadenine	2		

ERNDIM Response:

- SAICAR is very costly, however this will be reviewed periodically as other changes to the scheme may make this a viable addition in the future.
- Analytes only requested by two participants are not yet in demand enough to be added.

Q.7: Pterins – Urine (2 responses, 0.9% of all respondents)

Suggested Analytes to be added		Suggested Analytes to be removed
Total suggested = 1		Total suggested = 0
All analytes suggested		**All Analytes suggested**
Sepiapterin	2	

ERNDIM Response:

- Sepiaterin may be considered in the future, although currently there are very low numbers requesting this analyte.

Q.8: Quantitative amino acids (22 responses, 10.1% of all respondents)

Suggested Analytes to be added		Suggested Analytes to be removed	
Total suggested = 8		Total suggested = 7	
Analytes with >1 response		**Analytes with >1 response**	
Homocystine	7	2-Aminobutyric acid	2
Beta-alanine	2	Homocitrulline	3
Phosphoethanolamine	2	Sarcosine	2
Homocysteine	2	Sulphocystine	2

ERNDIM Response:

- The addition of phosphoethanolamine was tested in 2016 but it was not stable enough to include.
- Homocystine would also raise stability concerns.
- Beta-alanine is requested by too few participants to be included at this time.
- Too few participants have requested removal of any analytes. In particular sulphocystine would not be removed as it is considered to be a key diagnostic metabolite.

Q.9: Quantitative organic acids (21 responses, 9.7% of all respondents)

Suggested Analytes to be added		Suggested Analytes to be removed	
Total suggested = 26		Total suggested = 2	
Analytes with >1 response		**All Analytes suggested**	
Orotic acid	6	Mevalonic	1
Lactic acid	5	Vanillactic acid	1
2-methylbutirylglycine	2		
Glycolic acid	3		
Citric acid	2		
Propionylglycine	2		

ERNDIM Response:

- Orotic acid, Glycolic and lactic acid are included in the Special Assays in urine scheme.
- There were not enough requests for addition or removal of any other analyte to justify changes.

Q.10: Special assays – Dried Blood Spots (9 responses, 4.1% of all respondents)

Suggested Analytes to be added		Suggested Analytes to be removed	
Total suggested = 17		Total suggested = 2	
Analytes with >1 response		**All Analytes suggested**	
LysoGb1	2	Allo-isoleucine	1
LysoGb3	2	Homocysteine	1

ERNDIM Response:

- There are too few requests for the addition of LysoGb1 or LysoGb3.
- Allo-isoleucine and Homocysteine would not be considered for removal, as these are diagnostic metabolites relevant to newborn screening second tier tests and have limited alternative EQA.

Q.11: Special assays – serum (25 responses, 11.5% of all respondents)

Suggested Analytes to be added		Suggested Analytes to be removed	
Total suggested = 46		Total suggested = 7	
Analytes with >1 response		**All Analytes suggested**	
Acetoacetate	6	Coenzyme Q10	2
Biotinidase	3		
Campesterol	2		
Desmosterol	2		
Lathosterol	2		
Sitosterol	2		

ERNDIM Response:

Suggested additions

- Biotinidase - Enzyme activity, there is no commercially available analyte so it cannot be added. But is now included as a measurable analyte due to its presence in the sample matrix.
- Acetoacetate has previously been included in the scheme with some issues. 3-Hydroxybutyrate is included in the scheme and would be sufficient to identify the presence of ketonemia.
- Other requests were for too few analytes to be considered.
- The inclusion of Coenzyme Q10 has been requested over several years and is included in 2020 for the first time. The stability will be assessed at the end of the 2020 scheme year.

Q.12: Special assays – urine (24 responses, 11.1% of all respondents)

Suggested Analytes to be added		Suggested Analytes to be removed	
Total suggested = 45		Total suggested = 4	
Analytes with >1 response		**All Analytes suggested**	
Arabitol	2	Glycolic Acid	1
Dermatan sulfate	5	Lactic acid	1
Fructose	2	Sialic acid	1
Galactose	2	Sulphocysteine	1
Glutaric acid	2		
Heperan sulphate	3		
Keratan sulphate	3		
phosphoethanolamine	2		
sedoheptulose	2		

ERNDIM Response:

- The requests for addition of these analytes are too few to provide statistically relevant data if included. In addition some analytes are prohibitively expensive (e.g. Keratan sulphate).
- All analytes suggested for removal are measured and reported by a significant number of participants and therefore would not be removed at this time.

3.3. Do you have any other remarks, comments or suggestions for any of the schemes you subscribed to? (Q.13)

- Number of individual responses = 41 (= 9.8% of all responses).
- These comments are summarised under 3.10 (page 10) with the comments made in response to Q.22 (see 10).

3.4. Would your laboratory have an interest in participating in a new Quantitative pilot scheme similar to the special assays in serum scheme which would include Lysosphingolipids? (Q.14)

- A total of 206/217 respondents (94.9%) answered this question.
- The response options were 'Yes' (20/206, 9.7%), 'No' (177/206, 85.9%) or 'Yes if the following analytes were included…' (9/206, 4.4%).
- Labs that selected 'Yes if the following analytes were included…' were asked to provide a list of analytes they would like to be included. The responses are summarised below:

All Analytes (16)			
7alphaC4	1	LysoGm1	3
12alphaC4	1	LysoGM1ganglioside	1
7-KC	1	LysoGM2	2
COT	1	Lysohexosylceramide	1
Galactosylsphingosine	1	Lyso-SM	3
Lyso Gb3	4	Lyso-SM-509	1
Lyso-Gb1	1	Lysosphingolipids	1
Lysoglobotriaosylceramide	1	Lysosphingomyelin	1

3.5. Is your laboratory currently providing Metabolomics panels for diagnostic purposes? (Q.15)

No, we do not have any Metabolomics panels in use or development	151/ 206 (73.3%)
We are currently developing a Metabolomics panel for diagnostic use	25/206 (12.1%)
Yes, we offer a diagnostic Metabolomics panel	13/206 (6.3%)
We have a Metabolomics panel available but for research purposes only	17/206 (8.3%)

3.6. Would your laboratory be interested in participating in a Metabolomics pilot scheme? (Q.16)

Yes	36/206 (17.5%)
No	94/206 (45.6%)
Not yet, perhaps in 2 or more years	58/206 (28.2%)
Not yet, perhaps in 5 or more years	18/206 (8.7%)

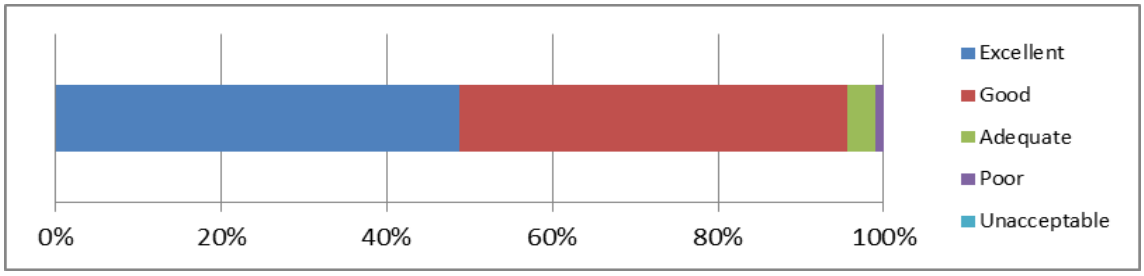
3.7. If you are interested in participating in a Metabolomics pilot scheme what sample type would be of most interest to you? (Q.17)

Plasma	43/91 (47.3%)
Urine	32/91 (35.2%)
Other (please specify)	16/91 (17.6%)

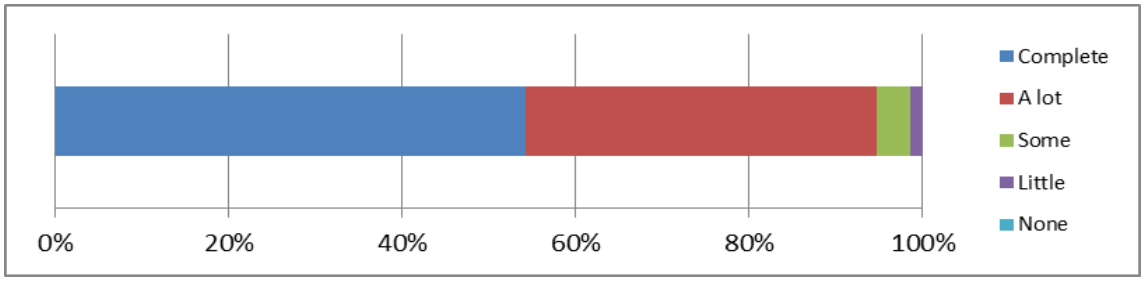
3.8. Comments on the overall performance of ERNDIM (Q.18 – 21)

- 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 95% 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.18: Overall, how do you rate the quality of products and services we provide?
(205 individual responses, 94.5% of all responses for this section)

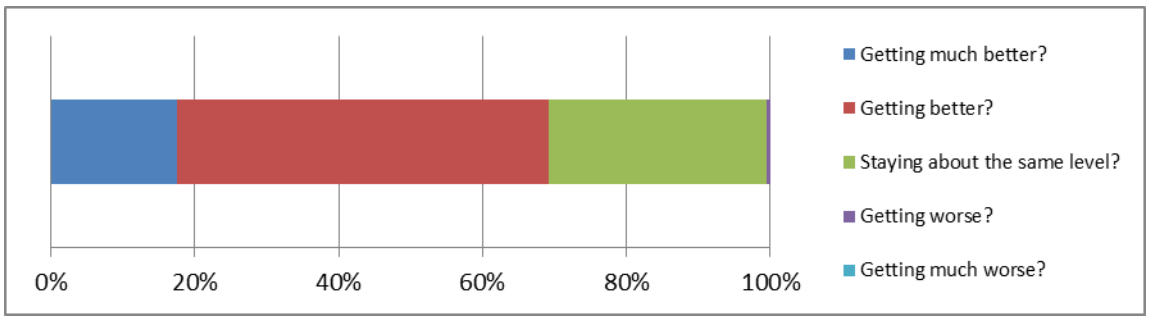


Q.19: What level of confidence do you have in us to deliver the products and services that you require? (205 individual responses, 94.5% of all responses for this section)



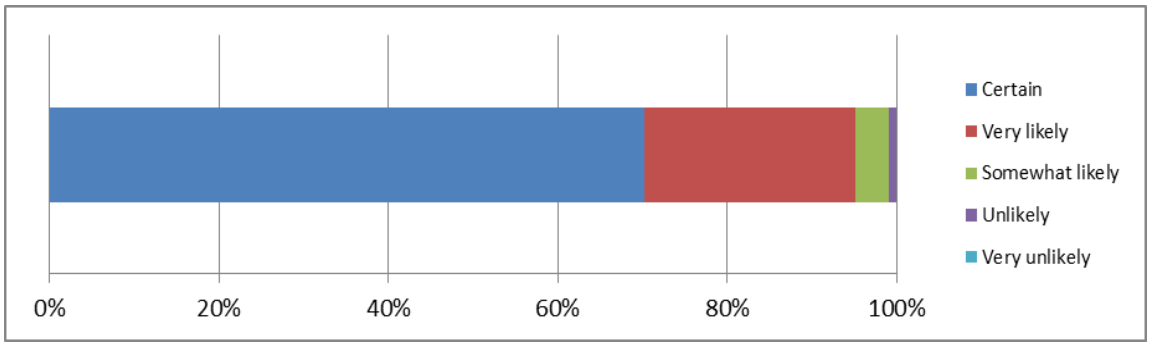
Q.20: Overall, is our performance...

(205 individual responses, 94.5% of all responses for this section)



Q.21: Based on our performance, how likely is it that you will use us in the future?

(205 individual responses, 94.5% of all responses)



3.9. Do you have any other remarks, comments or suggestions for how we could improve the services we provide? (Q.22)

- Number of individual responses = 52 (= 12.5% of all responses).
- These comments are summarised below with the comments made in response to Q22.

3.10. Summary of Remarks, comments or suggestions for improvements (Q.13 & Q.22)

- Total number of responses was 93 from 65 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	
• Communications: It has been great receiving reminders. In relation to performance, it may be helpful for the Quality Manager to be also emailed messages in relation to annual reports and/or performance related issues. Currently the QM doesn't appear to be contacted, even though we provide their contact details upon enrolment.	• The registration page allows 3 active contacts to be listed, these must be 3 unique contacts. Of these only the Primary and Secondary contacts will be routinely contacted and the Head of Lab/Quality Manager will only be contacted under specific circumstances, such as escalation of performance issues or difficulties contacting the Primary and Secondary contact. If you wish to update your contact information so that the Quality Manager receives all correspondence then please contact admin@erndim.org, however you would then need to provide a different Head of Lab/Quality Manager contact.
• If it is possible to increase the volume of sample urine/plasma being distributed to enable us to do further validation in relation to our participation to ERNDIM proficiency tests.	• EQA materials are not specifically designed for validation and the volume provided is intended to be sufficient for participation only. In order to increase the volume an additional cost or limits on the number of participants would have to been introduced. Control materials for some schemes can be purchased from MCA laboratories. Additionally if spare materials are available at the end of a scheme year these may be requested by participants.
• Please stop sending out frequent reminders to pay the yearly invoice in Jan / Feb when it isn't actually due til April. Our finance dept isn't going to pay until the due date so reminders are just an irritation.	• For some participants who have experienced email receipt problems or have missed previous reminders these emails are very important. Although the payment deadline is in early April payment can be made earlier and once received no further payment reminders will be sent.

Participant Comment	ERNDIM Response
2. EQA Schemes	
2.1. General	
• It would be useful to be able to obtain repeat samples more easily when trying to solve problems with assays.	• Where material is available ERNDIM is able to provide this to participants following completion of the scheme year or following the submission deadline for the specific sample. Please contact admin@erndim.org or visit the 'participant info' page of the registration website for further details.
• Report format for quantitative schemes is not very easy to follow, needs some explanation/guidance. Also, requires referring to website for more information on each analyte.	• Due to the number of analytes included in our Quantitative schemes it is not possible to review these individually within the annual report. We therefore recommend viewing the Scheme annual report alongside the individual lab annual reports and other report features available on the results submission website. The administration office can be contacted in the event of any unclear information and will be happy to assist.
• The ERNDIM DPT session at the SSIEM is excellent. It would be good to have additional sessions to discuss other qualitative EQA e.g. organic acids and acylcarnitines.	• This is something that the ERNDIM Scientific Advisory Board is giving some thought to. However it was noted that a session for the LEFB scheme was held at the September 2019 meeting and had poor attendance.
2.2. Website reporting	
• For qualitative acids organics, proficiency and acylcarnitines in DBS, clinical information should be available earlier and away from the deadline to submit the report of the analysis.	• Clinical information for these schemes are published on the results website before the submission deadlines for the relevant samples. We will make this clearer in the 2021 scheme instructions.
• The websites for reporting could be improved. They are often quite slow. The DPT scheme in particular feels like it should be modernised.	• The results submission website will continue to be developed and if you have any specific suggestions for improvements please contact admin@erndim.org. However at this time the priority is to complete work on the automated production of interim and annual reports in order to improve the time between submission of results and publication.
2.3. Acylcarnitines in DBS	
• The feedback of the DBS report should faster than it is now.	• Result submissions are currently assessed manually by our Scientific Advisors, this is a very labour intensive task and our Scientific Advisors have full time posts in diagnostic roles in addition to their ERNDIM contributions. We hope that the introduction of online reporting and subsequently assessment will reduce this workload and allow for a quicker publishing of results. Diagnoses will be circulated during 2020 in order to provide some feedback to participants in a timely manner.
2.4. CDG scheme	
• Increase the sample volume for CDG programme.	• The samples provided are from real patient samples, as such there is a limited amount of material. Although we are aware a proportion of participants require a larger volume, the 25ul sample volume provided is sufficient for the majority of participants. It is possible for participants requiring larger volumes to purchase additional material at a discounted fee. Please contact admin@erndim.org for further details.
Lysosomal Enzymes in fibroblasts scheme	
• Regarding the first sample of the year that is to be used as normal control for samples 02-06; it would be good to have enough material to run all enzymes again when samples 4-6 are analysed. This is not possible today so any problem in the measurement of sample 01 will affect all samples that year. We had a problem with protein concentration result and did not know it until reports came out. then it was too late to reanalyse samples 4-6.	• This comment will be passed to the Scientific Advisor. Due to the amount of material required, samples are cultured far in advance of sample distribution, therefore it may not be possible to make changes for the upcoming scheme year but these comments will be considered when planning future schemes.

Participant Comment	ERNDIM Response
• We suggest a change in ERNDIM Lysosomal Enzymes (fibroblasts) QA Scheme: - instead of 2 submission deadlines of 3 samples each, we suggest 3 submission deadlines of 2 samples each.	• Prior to the 2020 scheme year there were 4 submission deadlines for the LEFB scheme. This was adjusted to 2 deadlines for the 2020 scheme year, feedback will be considered and if a number of participants would prefer a greater number of submission deadlines this will be reviewed again.
2.5. Qualitative Organic Acids	
• In some cases, the concentration of abnormal metabolites was not high enough, or on borderline, and it was difficult to make a final judge. More detailed clinical information would be of helpful in such cases.	• ERNDIM recognises that some samples may be quite challenging for participants, however these can be of high educational value. In instances where samples are extremely challenging and there is poor proficiency as a result the sample may be retrospectively designated as an educational sample in order to avoid penalising participants.
2.6. Quantitative Amino Acids	
• We would prefer it if the 8 samples for Amino Acids and special assays could be spaced out more evening over the year, rather than monthly from March -October. We currently have 4 months of the year with no EQA for our metabolic assays which is not ideal.	• At this time it would be logistically very difficult for us to spread the submission deadlines more evenly through the year.
2.7. Special Assays in DBS	
• DBS samples are insufficient to measure all analytes. In addition, in clinical practice for monitoring we test all DBS samples in duplicate so we cannot test as if clinical samples	• Due to the limited availability of bulk material it is not possible at this time to increase the individual sample sizes prepared. Where possible, we do allow participants to request registration for a second set of samples if they perform a method requiring more sample.
2.8. Special Assays in serum	
• Is it possible to change the concentration of NEFA in SAS sample?	• NEFA is present in the sample matrix and is not added as a spiked analyte so it is not possible to vary it's concentration in the samples..
2.9. Urine Mucoploysaccharides	
• The MPS qualitative scheme currently asks for 'total MPS' (value) and qualitative assessment of whether dermatan, heparan, chondroitin and keratan sulfates are normal or abnormal. However, our lab no longer uses total MPS by a traditional method as a first-line test - instead we use LC-MS/MS as a first line test, generating quantitative results for HS, DS and CS. More and more labs are likewise changing their approaches. It would therefore be useful to have the option of submitting quantitative results for each of the individual GAGs - either as part of the MPS scheme or within the special assays in urine scheme.	• The Scientific Advisor for the UMPS scheme is investigating the possibility of including additional fields in the future to allow participants to record measurements for the individual metabolites.

3.11. Please complete your name and institute address details (Q.23)

- Number of individual responses = 135 (= 33% of all responses).