ERNDIM

QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

2015 Participant Survey Report:

[2014 scheme year]

ERNDIM Administration Office
Manchester Centre for Genomic Medicine
6th floor, St Mary's Hospital
Oxford Road, Manchester
M13 9WL, UK
Tel: +44 161 276 6741
Fax: +44 161 850 1145
E-mail: admin@erndim.org
Web: www.erndim.org

1. Introduction

- Participants (565 contacts from 362 centres) were sent the link to the ERNDIM Participant Survey on the Survey Monkey website (www.surveymonkey.com) on 21st May 2015. We asked participants to answer questions relating to the 2014 EQA schemes. The closing date for the survey was 10th July 2015.

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.
- Over 50% of the laboratories that participated in the 2014 schemes responded to the survey with the response rate for each of the schemes being between 53-64%.
- The survey has again highlighted areas where we need to improve such as the lack of website reporting for all of the qualitative schemes and low sample volume for some of the qualitative schemes. However It is gratifying to see that 38.5% of respondents rate the quality of products and services we provide as excellent and that the majority 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.
- We are working towards moving all the qualitative schemes to website reporting with the current aim being to test website reporting for the CDG and Qualitative Organic Acids scheme in 2016, with the Acylcarnitines in DBS scheme moving to website reporting in 2017/18.
- 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 and a recent appeal to members of the Society for the Study of Inborn Errors of Metabolism (SSIME, www.ssiem.org) led to a number of laboratories contacting us regarding possible sample donations.
- 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, which starts on page 9, where we answer some of your comments, interesting and we would welcome any other comments or suggestions for improvements.

3. Survey Responses

- 252/565 contacts from 219/362 centres in 52 countries responded to the survey. The response rate by centre was 58.6% (compared to 51.9% in the last survey) and the individual response rate was 34.4% (compared to 35.6% in the last survey).

Question 1: Please rate the following aspects for each of the ERNDIM quality assurance schemes that you subscribe to

- Number of centre responses = 217 centres (= 99% of all responses)
- 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 Diagnostic Proficiency Schemes (64% of 2014 scheme participants) and the lowest was for Cystine in WBC (51% of 2014 scheme participants). The response rate for 9 of the 12 EQA schemes was higher than in the 2014 survey (= 2013 scheme year, Figure 1).

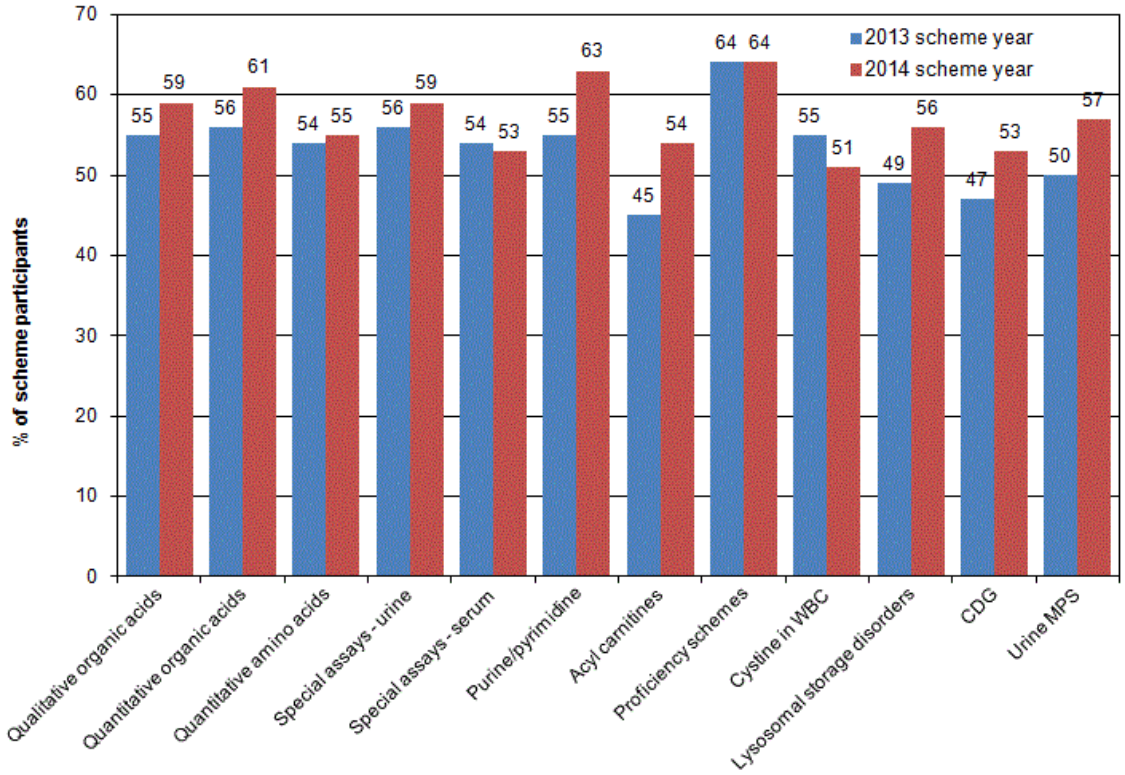


Figure 1. Survey responses per EQA scheme (Question 1) as a percentage of the EQA scheme participants

- Participants were asked to rate the following aspects of each scheme:
  1. Frequency of samples
  2. Sample volume
  3. Appropriateness of analyte concentration
  4. Adequacy of the report
  5. Website display
  6. Usefulness of the annual report
  7. Value for money
  8. 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
- Scores ≤ 1.5 are highlighted in blue and scores ≥ 2.0 are highlighted in red.

Table 1. Average scores per scheme (Question 1)

	Average Scores (survey year)							
EQA Scheme	**2015**	**2014**	**2013**	**2012**	**2011**	**2007**	**2004**	**2001**
All schemes	**1.8**	**1.7**	**1.7**	**1.7**	**1.8**	**1.7**	**2.0**	**2.0**
Qualitative organic acids	1.7	1.7	1.7	1.7	1.7	1.6	2.0	1.9
Quantitative organic acids	1.8	1.7	1.7	1.7	1.9	1.7	1.9	2.1
Quantitative amino acids	1.7	1.7	1.7	1.7	1.8	1.7	1.9	2.0
Special assays - urine	1.7	1.7	1.7	1.7	1.7	1.8	1.9	2.1
Special assays - serum	1.7	1.7	1.7	1.7	1.8	1.7	1.8	2.0
Purines & pyrimidines	1.8	1.7	1.7	1.7	1.9	1.6	1.8	2.1
Acylcarnitines in DBS	1.9	2.0	1.9	1.9	2.0	2.0	2.3	-
Diagnostic Proficiency Testing	1.7	1.7	1.7	1.8	1.8	1.7	2.0	2.0
Cystine in white blood cells	1.8	1.8	1.6	1.7	1.6	1.4	-	-
Lysosomal storage enzymes (fibroblasts)	1.9	2.0	1.9	2.0	2.1	-	-	-
Congenital disorders of glycosylation	2.0	2.0	1.9	1.8	1.9	-	-	-
Urine Mucopolysaccharides	1.8	1.8	1.8	1.8	-	-	-	-

- The average scores per scheme since 2001 are shown in Table 1 and Figure 2.
- The overall score for all aspects of all schemes was 1.8, which is slightly higher than in 2014 (1.7). Eight of the EQA schemes had the same score as last year, 2 schemes had worse scores than last year (Quant Organic Acids and Purines & Pyrimidines) and 2 schemes had better scores (Acylcarnitines and Lysosomal Storage Enzymes).
- The best scoring schemes were Qualitative Organic Acids, Quantitative Amino Acids, Special Assays in serum and urine, and the DPT scheme which all scored 1.7. The worst scoring scheme was CDG which scored 2.0.
- The scores for each scheme in each of the individual aspects are given in table 2. The score for 4 out of the 8 of the individual aspects have improved or stayed the same since last year, while 'Frequency of samples', 'Usefulness of the annual report', 'Value for money' and 'Billing arrangements' all have a worse score than last year.
- The worst scoring aspects were 'Value for money' and 'Billing arrangements' which both scored 1.9; with the best scoring aspects being 'Frequency of samples', 'Appropriateness of analyte concentration' and 'Adequacy of the report' (all scored 1.7).
- The score for 'Sample volume' has remained the same as in 2014 (1.8) however three schemes still scored 2.0 or more (LSDs = 2.1, CDG = 2.5, Urine MPS = 2.0) for this aspect. For the second year the 'Sample volume' score for CDG, was the worst score in the survey, although it has slightly improved since 2014 (2.5 in 2015 compared to 2.6 in 2014).
- Website display' had the most schemes (= 4) with scores over 2.0. Three of these schemes (Qual Organic Acids = 2.0, Acylcarnitines = 2.3, CDG = 2.2) do not yet have online results submission but Cystine in WBC (= 2.0) is on the SKML results website.
- The best scores of the whole survey (all 1.5) were for 'Frequency of samples' (Cystine in WBC), 'Sample volume' (Purines and pyrimidines) and 'Adequacy of the report (Qual Organic Acids).
- The most improved scores of the whole survey were for Cystine in WBC (frequency of samples, 1.5 compared to 1.7 in 2014; sample volume, 1.6 compared to 1.9 in 2014; appropriateness of analyte concentration, 1.7 compared to 1.9 in 2014) and Lysosomal Enzymes (sample volume, 2.1 compared to 2.3 in 2014).

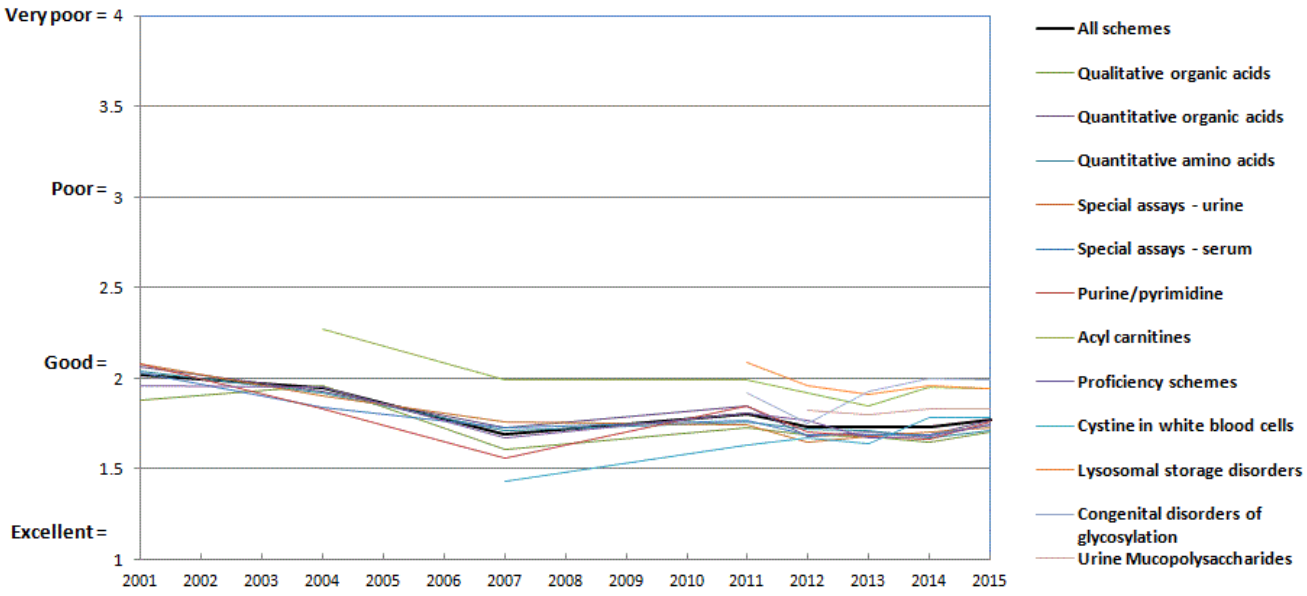


Figure 2. Average score per EQA scheme (Question 1)

Table 2: Average scores per aspect of each scheme (Question 1)

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)
Qual. organic acids	1.7	1.8	1.6	1.5	2.0	1.6	1.8	1.8	**1.7**	104 (55.0%)
Quant. organic acids	1.6	1.7	1.8	1.7	1.8	1.8	1.9	1.9	**1.8**	62 (55.9%)
Quant. amino acids	1.6	1.6	1.7	1.7	1.7	1.7	1.9	1.9	**1.7**	136 (53.8%)
Special assays - urine	1.6	1.8	1.7	1.7	1.7	1.8	1.9	1.8	**1.7**	93 (56.0%)
Special assays - serum	1.6	1.6	1.8	1.8	1.7	1.8	1.8	1.9	**1.7**	114 (54.3%)
Purines/pyrimidines	1.6	1.5	1.8	1.8	1.8	1.9	1.9	1.8	**1.8**	31 (55.4%)
Acyl carnitines	2.2	1.9	1.8	1.9	2.3	1.8	2.0	1.9	**1.9**	55 (45.1%)
Proficiency schemes	1.7	1.9	1.7	1.6	1.9	1.6	1.7	1.8	**1.7**	66 (63.5%)
Cystine in WBC	1.5	1.6	1.7	2.0	2.0	1.7	1.8	2.0	**1.8**	18 (54.5%)
LSD	1.7	2.1	1.9	2.1	1.9	2.0	2.0	1.9	**1.9**	35 (48.6%)
CDG	1.8	2.5	1.9	1.8	2.2	1.8	2.1	2.0	**2.0**	28 (46.7%)
Urine MPS	1.7	2.0	1.8	1.8	1.9	1.8	1.8	1.8	**1.8**	52 (50.0%)
Average for all schemes	**1.7**	**1.8**	**1.7**	**1.7**	**1.8**	**1.8**	**1.9**	**1.9**		

Questions 2 to 7: Analytes in Quantitative Schemes

- A total of 91/252 individuals (36%) 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 and 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.

Q.2: Quantitative amino acids (46 responses, 18.3% of all respondents)

Suggested Analytes to be added

Total suggested = 22

Analytes with >1 response

arginosuccinic acid (ASA)	n = 10
tryptophan	n = 7
homocystine	n = 6
sulfocysteine	n = 6
alloisoleucine	n = 5
3-methylhistidine	n = 4
5-hydroxylysine	n = 3
cystathionine	n = 2
Pipecolic acid	n = 2

Suggested Analytes to be removed

Total suggested = 9

Analytes with >1 response

saccharopine	n = 10
homocitruliine	n = 8
2-aminobutyric acid	n = 4
phosphoethanolamine	n = 4
alloisoleucine	n = 3
asparagine	n = 2

Q.3: Quantitative organic acids (26 responses, 10.3% of all respondents)

Suggested Analytes to be added		Suggested Analytes to be removed	
Total suggested = 21		Total suggested = 5	
Analytes with >1 response		**All Analytes suggested**	
3-hydroxy-glutaric acid	n = 9	3-hydroxyisobutyric acid	n = 2
suberylglycine	n = 6	5-hydroxyindolic acid	n = 1
3-methylglutaconic	n = 5	DL-glyceric	n = 1
methyl citrate	n = 4	keto glutaric	n = 1
3-hydroxybutyric acid	n = 3	vanillactic	n = 1
n-acetylaspartic acid	n = 3		
homogentisic acid	n = 2		
isovalerylglycine	n = 2		
lactic acid	n = 2		
succinylacetone	n = 2		

ERNDIM Response:

- 3-hydroxyisobutyric acid and malic acid will be removed from the 2016 scheme onwards, with glycolic acid being removed for the 2017 scheme onwards.
- 3-methylcitric acid (feasibility study), 3-hydroxyglutaric acid, 3-methylglutaconic acid and N-acetylaspartic acid will be added to the 2016 scheme.

Q.4: Purines & pyrimidines (6 responses, 2.4% of all respondents)

Suggested Analytes to be added		Suggested Analytes to be removed	
Total suggested = 5		Total suggested = 3	
Analytes with >1 response		**All Analytes suggested**	
SAICAR	n = 2	Deoxy-guanosine	n = 1
Succinyladenosine (SAdo)	n = 2	Deoxy-inosine	n = 1
		S-adenosine	n = 1

ERNDIM Response:

- The additions of SAICAR (SAICAriboside) and SAdo to this scheme have been requested in the past. The availability of both these analytes is very limited and unfortunately the addition of these compounds is financially not feasible.

Q.5: Lysosomal Enzymes (25 responses, 9.9% of all respondents)

Suggested Analytes to be added		Suggested Analytes to be removed	
Total suggested = 20		Total suggested = 4	
Analytes with >1 response		**All Analytes suggested**	
Arylsulfatase A	n = 6	ß-galactocerebrosidase	n = 4
iduronate sulphatase	n = 5	Sphingomyelinase	n = 2
Arylsulphatase B	n = 4	Aryl sulfatase A	n = 1
alpha-L iduronidase	n = 2	Galactose-6-sulphate	n = 1
beta-glucuronidase	n = 2		
Lysosomal enzymes in DBS (GALC, GAA, GLA, ASM, ABG)	n = 2		
MPS III enzymes	n = 2		

ERNDIM Response:

- It is not possible to restart the Lysosomal Enzymes in DBS pilot scheme due to a lack of suitable clinical materials to use as the EQA materials (also discussed under 'suggestions for future schemes, page12).

Q.6: Special assays – serum (33 responses, 13.1% of all respondents)

Suggested Analytes to be added		Suggested Analytes to be removed
Total suggested = 39		Total suggested = 0
Analytes with >1 response		**All Analytes suggested**
biotinidase	n = 6	NONE
acetoacetate	n = 3	
Cholesterol	n = 3	
Increased range of acylcarnitines	n = 3	
more Free Fatty Acids/wider range of concentrations	n = 3	
Total carnitine	n = 3	
3-methylglutaconic acid	n = 2	
asymmetric dimethylarginine	n = 2	
C2, C3, C4,C5,C6,C10 & C14:1 Carnitines	n = 2	
Glutaric Acid	n = 2	

ERNDIM Response:

- Biotinidase is not commercially available. CDC (www.cdc.gov/nsqap) provides an EQA scheme for biotidinase in dried blood spots.
- Acetoacetate is a very unstable analyte. It was added in the first years of running this scheme (until 2003) with very poor results (low recovery, bad precision and linearity), and it was decided to stop the addition.
- The Scientific Advisory Board (SAB) meeting in November 2015 decided that Cholesterol will not be added to this scheme but it will be possible to submit results for comparison between labs.
- C3 & C5 carnitines have already been added to the 2016 scheme.
- C14:1 carnitine is not commercially available.
- C2, C4, C6 & C10 carntines will not be added to this scheme as the ERNDIM is planning to introduce a Quantitative Acylcarnitines scheme in 2017.
- Total carnitines are not commercially available so cannot be added to the scheme.
- NEFA are not added to the samples for this scheme but are present in the sample matrix however results can be submitted for comparison between labs.
- The SAB meeting in November 2015 rejected the addition of 3-methylglutaconic acid, glutaric acid and dimethylarginine as these are not important analytes for the diagnosis of inborn errors or metabolism

Q.7: Special assays – urine (22 responses, 8.7% of all respondents)

Suggested Analytes to be added		Suggested Analytes to be removed
Total suggested = 34		Total suggested = 0
Analytes with >1 response		**All Analytes suggested**
cystine	n = 3	NONE
galactose	n = 3	
VMA	n = 3	
arabitol	n = 2	
carnitine total	n = 2	
fructose	n = 2	
ribitol	n = 2	

ERNDIM Response:

- The feasibility of adding cystine to the scheme will be investigated however the last meeting of the Scientific Advisory Board (SAB) in November 2015 rejected the addition of galactose.
- The possible addition of vanillylmandelic acid (VMA), arabitol, fructose, and ribitol was discussed by the SAB in November 2014 and rejected.
- Total carnitines are not commercially available so cannot be added to the scheme.

Question 8: ERNDIM is investigating the possibility of a Quantitative Acylcarnitines in serum EQA scheme. Would your laboratory be interested in participating in such a scheme?

- Number of centres responses = 210 (= 83% of all responses).
- 102 centres answered 'Yes, they would be interested in participating in a Quantitative Acylcarnitines in serum EQA scheme' (= 49% of centres that answered this question).

ERNDIM Response

- We are currently investigating the possibility of setting up Quantitative Acylcarnitines in serum EQA scheme which would be organised in a similar way to the Special Assays schemes.

Question 9: Do you have any other remarks, comments or suggestions for schemes you subscribed to?

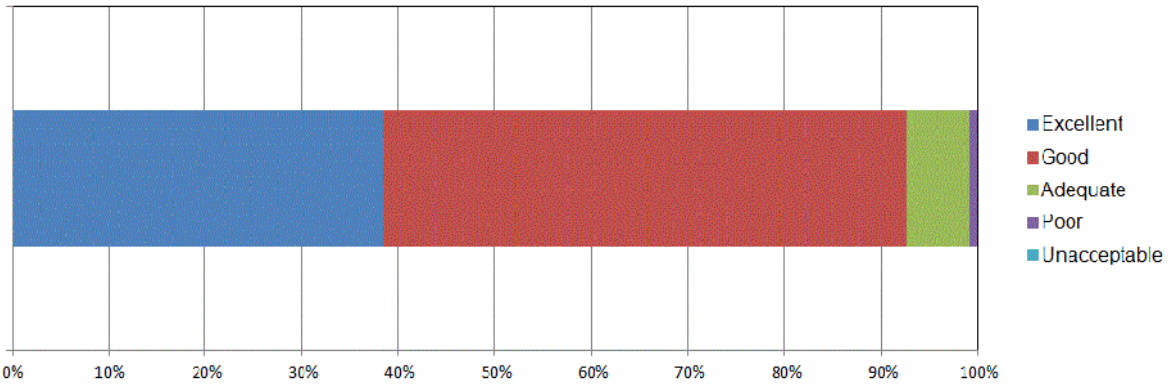
- Number of individual responses = 59 (= 23% of all responses)
- These comments are summarised with the comments made in response to Q14 on page 9.

Questions 10 to 13: Comments on the overall performance of ERNDIM

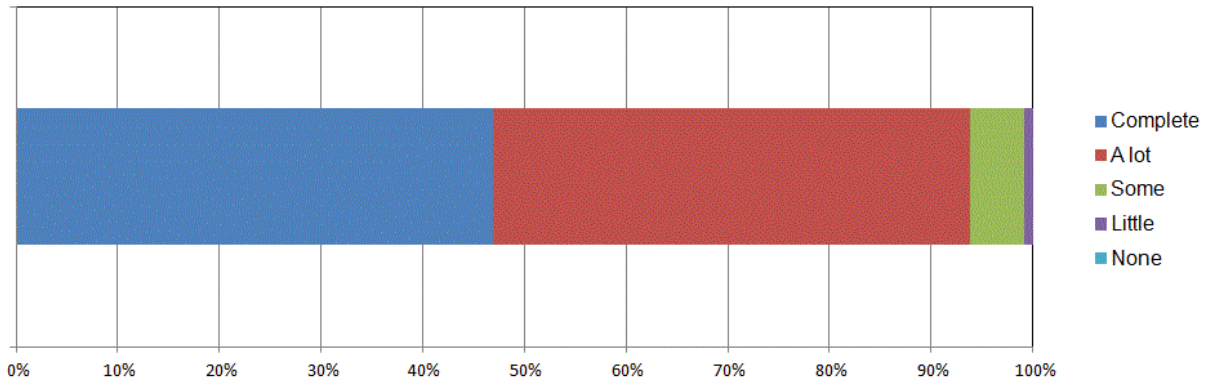
- This aim of this section is to assess participants' perception of the overall performance of ERNDIM.
- In summary, 93% of respondents rated the quality of services provided by ERNDIM as 'excellent' or 'good'; with 94% of respondents having 'complete' or 'a lot' of confidence that ERNDIM can deliver the service required by participants.
- 73% 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.10: Overall, how do you rate the quality of products and services we provide?

(244 individual responses, 97% of all responses)

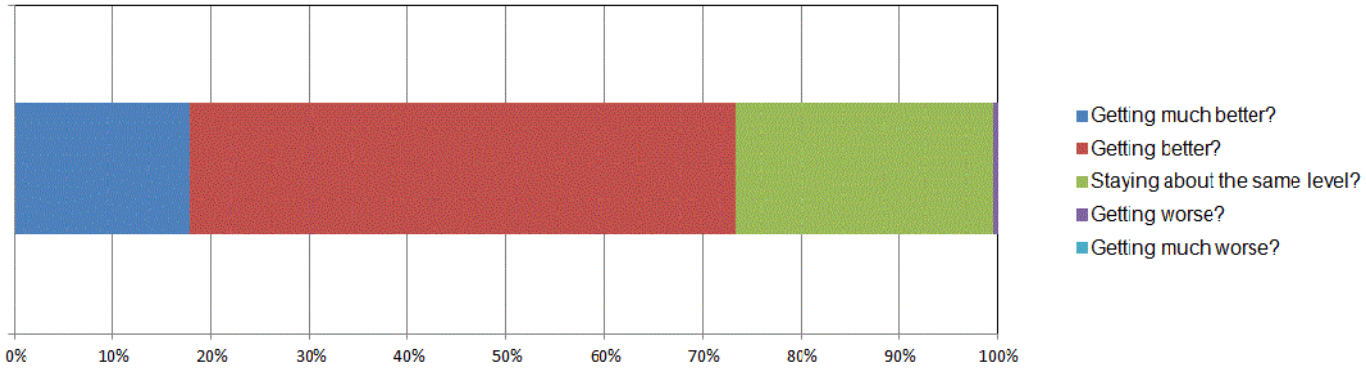


Q.11: What level of confidence do you have in us to deliver the products and services that you require? (241 individual responses, 96% of all responses)



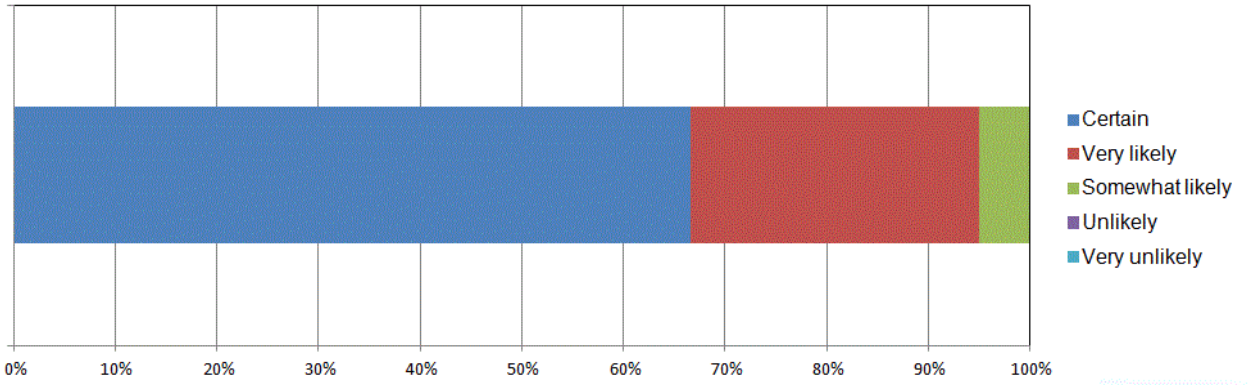
Q.12: Overall, is our performance...

(240 individual responses, 95% of all responses)



Q.13: Based on our performance, how likely is it that you will use us in the future?

(243 individual responses, 96% of all responses)



Question 14: Do you have any other remarks, comments or suggestions for how we could improve the services we provide?

- Number of individual responses = 67 (= 27% of all responses)
- These comments are summarised below with the comments made in response to Q9.

Questions 9 & 14: Remarks, comments or suggestions for improvements

- Total number of responses was 126 from 97 individuals (= 38% 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	
• The cost of the schemes increases each year.	• ERNDIM is a not-for-profit foundation and the prices of the EQA schemes reflect the cost of the schemes and the costs of running the organisation as we make progress towards applying for accreditation.
• Would it be possible to introduce credit card payments?	• We will investigate if this is possible without increasing costs to participants.
2. EQA Schemes	
2.1. General	
• It would be helpful to obtain more information about the recommended analytical procedures for the analytes in the EQA scheme.	• There are some method documents on www.erndim.org under 'Training' but many of these need to be updated. We will look at updating the existing documents and adding some new methods.

Participant Comment	ERNDIM Response
• There were, again, a number of comments on the frequency of submission deadlines – some wanting more frequent (up to 12 per year) and some wanting less frequent deadlines (4 per year).	• Increasing the number of submission deadlines so that there were 12 submissions per year would make running the schemes extremely difficult. • Decreasing the number of submissions per year would mean very long periods without EQA coverage which would not be acceptable. • There are therefore no plans at the moment to alter the number or frequency of submission deadlines for the EQA schemes.
• Send samples monthly instead of in one parcel.	• The samples are sent in one parcel at the beginning of the scheme year to reduce costs.
• Increased number of sample distributions for some of the qualitative schemes.	• For the qualitative schemes there are often problems sourcing suitable clinical material of a sufficient volume to use as the EQA materials. For most of these schemes the Scientific Advisor sources all the EQA materials themselves and we would welcome offers to donate suitable samples from participating centres. Please contact the Administration office if you would be interested in donating a sample.
• Lack of website reporting for all the qualitative schemes.	• ERNDIM's long term aim is to move all of the qualitative schemes to the CSCQ Results website. It is planned that the CDG and the Qualitative Organic Acids schemes will begin to move to website reporting in 2016 with the Acylcarnitines in DBS scheme moving to website reporting in 2017/18.
• Faster access to the annual reports.	• We agree that the annual reports need to be published earlier. Currently the final results for each scheme are ratified by the Scientific Advisory Board at its spring meeting after the end of the scheme year and all annual reports are published as soon as possible after that meeting. However we are working on a revised calendar for 2017 which should allow the annual reports for some schemes to be published earlier.
• Certificates of Participation to be sent earlier and notification to be sent to labs when they are released.	• In the past the Certificates have sometimes been published in August which, we agree, is too late. The delay is because all the scheme results need to be finalised before any of the certificates can be produced. In 2014 and 2015 the certificates were published in July. This year we're hoping to publish them in June and we're working towards publishing them earlier next year. • In 2015, for the first time the certificates were available for labs to download from the www.erdimqa.nl website. All labs were sent a notification email when the certificates were available.
2.2. Acylcarnitines in DBS	
• Delivery of samples is delayed.	• The EQA materials for the scheme are real clinical samples and delays in sample dispatch are due to difficulties obtaining suitable samples.
• Interim reports should be published earlier.	• We are working towards online submission of results for this scheme which will make it easier for the Scientific Advisor to evaluate the results and send out reports earlier.
2.3. CDG scheme	
• Reports should be sent out in a timely fashion and an interim report after the first round should be provided.	• We agree that this needs to be improved. For the 2016 scheme the diagnoses for the samples in the first round will be sent to all participants at the end of April, and the diagnoses for the second round will be sent at the end of September.
• Low sample volume.	• This scheme uses real clinical samples and at least 3ml plasma is needed for each sample. The difficulty obtaining suitable samples of a large enough volume means that the volume per EQA sample is restricted. It is possible to order additional sample volume at a reduced fee but the availability of this is limited. The Scientific Advisor regularly appeals for sample donations but with few responses. Please contact the Administration office if you would be interested in donating a sample.
2.4. DPT scheme	
• It would be helpful to have the demographic info available at the same time the samples are received.	• This information will be added to the sample dispatch letters for the 2017 schemes.

Participant Comment	ERNDIM Response
• The deadline in late June is problematic.	• We understand this difficulty but it is difficult to move this deadline any earlier as then it would be too close to the first submission deadline however, as stated under item 2.1, the calendar for this scheme is being reviewed.
• Problems with the submission deadlines for the second DPT round and middle Qualitative Organic Acids round coinciding.	• In future we will try to avoid these being in the same week.
2.5. Lysosomal Enzymes in fibroblasts	
• Provide a normal control with the sample.	• For the 2016 scheme results for samples will be submitted in pairs with one sample being the control for the other.
• Include a method for each enzyme.	• The Scientific Advisor and the scheme organiser will investigate if it is possible to modify the results website so this can be included when the results are submitted.
• The annual reports are very late.	• In the past the annual reports for this scheme have been published very late. This was due to miscommunications and in future the annual reports for this scheme will be published in line with the timetable for the other EQA schemes.
• The amount of sample sent should be increased. It would be much easier to work with a protein concentration around 1,0 mg.	• It is very difficult to increase the amount of material sent due to the very long time which is already needed to culture enough fibroblasts for the amount of material which is currently sent to participants. The Scientific Advisor tests all samples before they are sent and performs the assays with a protein concentration of 1 mg/ml and has found that the amount of material in the samples is enough.
• Please don't give negative scores if we do not measured the relevant (abnormal) enzyme.	• The scoring system was changed for the 2015 scheme so that a for each enzyme a maximum of 2 points each could be scored for CV and diagnosis, giving a maximum total of 4 points. If results were not submitted for an enzyme no score was recorded. Full details of the scoring scheme can be found in the 2014 LSE in fibroblasts annual report (www.erndim.org under Meetings & Reports\EQA scheme annual reports).
2.6. Quantitative Amino Acids	
• The report could be clearer with more comments on results and interpretation.	• More comments have been added to the results for the 2015 schemes.
• The z score should be given.	• This will be included in the reports for the 2016 scheme.
2.7. Qualitative Organic Acids.	
• Interim reports should be published earlier.	• We are working towards online submission of results for this scheme which will make it easier for the Scientific Advisor to evaluate the results and send out reports earlier.
• Slow responses to email communication.	• We are sorry for any delays in replying to emails. These schemes do not yet have online results submission and the Scientific Advisors for each of these schemes have over 100 participants each and while they try to respond to all emails in a timely manner this is not always possible. If you have any problems contacting one of the Scientific Advisors please contact the Administration Office and we will direct your query to the correct person.
• Online submission of results and better data for result analysis e.g. method specific and std based evaluation.	• We are working on website submission of results and hope to begin implementing this in 2016.
• Problems with the submission deadlines for the second DPT round and middle Qualitative Organic Acids round coinciding.	• In future we will try to avoid these being in the same week.
2.8. Urine MPS	
• Thank you for moving the final distribution of the urine MPS to the end of the summer and not to coincide with the DPT & Qual OA schemes in June.	• We're pleased you have found this helpful.

Participant Comment	ERNDIM Response
• Improve the time from submitting results to receiving the report for urine MPS scheme.	• As stated under item 2.1, the calendar for this scheme is being reviewed which should allow the annual reports for this scheme to be published earlier for the 2016 scheme onwards.
• Scoring system is unfair and does not reflect best practice. It is not recommended to delineate the MPS subtype based on urine tests alone....In this scheme if you do not pinpoint the MPS subtype based on the urine findings you are only marked as partially correct.	• In metabolic screening it is best practice to make the differential diagnosis as narrow as possible based on the results of urine analysis. In urine MPS screening the type of GAG detected and the resulting differential diagnosis narrows down the number of enzyme tests required to establish diagnosis, probably lowering costs. The scoring criteria reflect the results that can be achieved by the majority of the labs and are approved by the ERNDIM Scientific Advisory Board. If anyone would like to discuss specific cases they are invited to email the Scientific Advisor for the Urine MPS scheme.
• The scoring system for urine MPS gives one mark for quantitative analysis per sample which means labs that only perform qualitative analysis are at a disadvantage in the scoring (only a maximum of 18 can be achieved in the year). Unless ERNDIM is suggesting that all labs should be performing and reporting total GAG concentration then the scoring system should be changed to account for this. Is there good evidence that not performing quantitative GAGs causes poorer performance?	• The scoring system for the Urine MPS scheme reflects the approach that most laboratories use in urine MPS screening: a quantitative first tier test to identify possible MPS patients and qualitative second tier test to confirm and specify abnormal GAG accumulation. The ERNDIM EQA scheme catalogue states that for the Urine MPS scheme quantitative (related to creatinine) and qualitative analysis of mucopolysaccharides is required with interpretation of the results obtained. So both analytical results are scored with one point.
3. Suggestions for future schemes	We do welcome suggestions for future schemes but unfortunately it is not possible to cater for every request.
• Acyl carnitines in serum.	• Plans for this scheme are being investigated.
• Cognitive scheme for amino acids.	• We are hoping to introduce a small pilot scheme for cognitive amino acids later this year. Initially participation will be limited but the aim will be to increase the number of participants once the pilot phase is completed.
• Reintroduce the Lysosomal Enzymes in DBS pilot scheme.	• Unfortunately it will not be possible to reintroduce this pilot scheme unless sufficient samples to run the scheme are donated by participating labs as there is a lack of suitable clinical materials to use as the EQA materials. Please contact the Administration office if you would be interested in donating a sample.

Question 14 Please complete your name and institute address details.

- Number of individual responses = 231 (= 92% of all responses).