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Diagnostic Proficiency Testing

Survey 2017 - Switzerland

Final Report

Date of issue: 22nd January 2018
Amended report issued: 23rd May 2018[1]

prepared by
Brian Fowler

Version Number (& Date)	Amendments
[1] Version 2 (23 May 2018)	• Page 20, Table 'Detailed Scored: A,B,C': total score for lab 4 has been changed from 8 to 12. Scores for individual samples were already correct and have not been changed.

1. Geographical distribution of participants

In 2017, 21 laboratories from 11 countries subscribed to the scheme. All laboratories submitted results for both sample batches.

Country	Number of participants
Australia	2
Austria	2
Canada	3
China	1
Estonia	1
France	1
Germany	3
Norway	1
Sweden	2
Switzerland	1
USA	4

2. Samples and Shipment

The samples have been heat-treated. They were pre-analysed in our institute after 3 days incubation at ambient temperature (to mimic possible changes that might arise during transport). In all six samples the typical metabolic profiles were preserved after this process. This year, in addition to the common sample and those originating from our own centre, **samples were donated by: K. Ounap, Tartu, Estonia; C.D. Langerhans, Heidelberg, Germany,** to whom we express our gratitude.

3. Tests

Ability to analyse amino acids, organic acids, mucopolysaccharides, oligosaccharides and purines/pyrimidines was required in 2017.

4. Schedule of the scheme in 2017

Sample distribution	Feb 1st 2017
Start of analysis of Survey 2017/1	Feb 20th, 2017
Survey 2017/1 - Results submission	March 13th, 2017
Survey 2017/1 - Reports on website	May 4th, 2017
Start of analysis of Survey 2017/2	May 22nd, 2017
Survey 2017/2 – Results submission	June 12th , 2017
Survey 2017/2 - Reports on website	July 31st , 2017
Annual meeting of participants	ERNDIM, Manchester, November 21, 2017
Annual Report 2017	December 2017

We continued to use the evaluation programme to generate individual lab reports and these were made available on the CSCQ website in good time close to the foreseen dates. Feedback on the content and style of these reports is invited.

5. Receipt of samples and results

Receipt of samples

The urine samples were distributed to participants on **February 6th** at ambient temperature by CSCQ using the courier DHL. Delivery times of samples reported by the courier were all within a maximum of four days although 2 labs reported receipt dates that were several days later, suggesting possible internal delays within the institution.
Specific details regarding your own sample are available on request.

Date of reporting of results

All labs returned results for both the first and second surveys and by the deadline.

6. Scoring system

Two criteria are evaluated: analytical performance, interpretative proficiency including recommendations for further investigations. Due to the large variability in reporting results in various countries, recommendations pertaining to treatment are not evaluated in proficiency testing, however they may be considered by the scheme organisers in evaluating interpretation.

A	Analytical performance	Correct results of the appropriate tests	2	max 2
		Partially correct or non-standard methods	1	
		Unsatisfactory or misleading	0	
I	Interpretative proficiency & Recommendations	Good (diagnosis was established)	2	max 2
		Helpful but incomplete	1	
		Misleading or wrong diagnosis	0	

The **total score** is calculated as a sum of these two criteria. The maximum to be achieved is 4 points per sample. The scores were calculated only for laboratories submitting results.

7. Results of samples and evaluation of reporting

Sample A: **Citrullinemia type I (argininosuccinate synthase deficiency), OMIM #603470**

This was the common sample that was distributed to all five DPT schemes**.**

Patient details provided
Infant presented at Emergency Department. Febrile, query infection. Sample collected after commencing therapy.

Further information
This was a sample taken from a patient who presented at A+E at 3 days of age, febrile - query infection. An ammonia performed on the first sample submitted to the lab gave a result of 542 µmol/L. On the first urine sample the orotic acid was 1800 µmol/mmol creatinine and the citrulline was 1400 µmol/mmol creatinine.

Analytical performance:
The finding of increased citrulline (21/21 labs) and of increased orotic acid (20/21 labs), each scored with one point was considered correct.

Interpretative proficiency
The correct diagnosis of citrullinaemia type 1, reported by 20 labs was considered correct (2 points). One lab reported this as the second possible diagnosis and received one point.

Recommendations:
Appropriate recommendations were:
Plasma ammonia; plasma amino acids;; mutation analysis / enzyme assay; specified emergency treatment.

Overall impression:
Very high proficiency of a straightforward sample.
Proficiency: Analytical 98%; Interpretative 98%; Overall 98%.

This sample was considered by the SAB to be eligible for critical error. .No lab qualified for this sample

Analytical Details

Creatinine
n=21
median= 0.61
mean= 0.69
SD= 0.21
min, max= [0.48, 2.70]

pH
n=10
median= 5.00
mean= 5.25
SD= 0.18
min, max= [5.00, 6.00]

Spot tests
Glucose: + (n=4)

Sample A: contd.

Amino Acid Analysis

	n	points
Citrulline increase	21	1

Citrulline
n=20
median= 11505.00
mean= 12338.75
SD= 6748.53
min, max= [129.34, 23468.30]

Arginine
n=9
median= 64.70
mean= 69.85
SD= 16.06
min, max= [50.00, 106.70]

Organic Acid, Purine/ Pyrimidine analysis

	n	points
Orotic acid	20	1

Orotic acid (Organic acids)
n=7
median= 75.00
mean= 65.10
SD= 25.88
min, max= [27.70, 106.00]

Orotic acid (Purine/Pyrimidines)
n=5
median= 222.00
mean= 254.00
SD= 178.03
min, max= [11.00, 465.00]

Interpretation

	n	Points
Citrullinaemia type 1	20	2
OTC and Citrullinaemia as second diagnosis (1)	1	1

Full details of the results of this common sample are to be found on the ERNDIM website with the following link:

http://www.erndim.org/store/docs/2017DPTcommonsample-Joan-HESUCABA615035-21-12-2017.pdf

Sample B: Mevalonic aciduria due to mevalonate kinase deficiency, OMIM #610377

Patient details provided:
Male child ill at 19 months with fever and cough, developmental delay, subsequently neurological abnormalities noticed. Urine collected at age of three on treatment..

Further information
Male child ill at 19 months with fever and cough (bacterial pneumonia diagnosed), increased ASAT, ALAT and CK. Subsequently regression in motor development, not walking, lack of head control. At 2y 3m muscular hypotonia, ataxia and tremor noticed. Suspicion of an autoimmune disorder led to treatment with prednisolone with positive effect. Gene panel testing revealed two heterozygous mutations of the MVK gene. Concurrent organic acid analysis revealed mevalonic aciduria (7203 mmol/mol creatinine). Urine collected at age of three on treatment.

Analytical performance*:*
The finding of increased mevalonic acid and/or mevalonolactone was key and scored with 2 points (21/21 labs).

Interpretative proficiency:
The correct diagnosis of mevalonate kinase deficiency was correct and scored with two points (21/21 labs).

Recommendations:
Appropriate ones given were:
Repeat MVA measurement (n=3); ule out MA-HIDS / IgD, IgA (n=8);
enzyme assay (n=10);
mutation analysis (n=19).

Overall impression
Excellent performance with 100% efficiency.

This was considered by the SAB to be eligible for critical error. No lab qualified for this sample

Analytical Details

Creatinine
n=20
median= 1.21
mean= 1.20
SD= 0
min, max= [1.07, 1.33]

pH
n=10
median= 6.00
mean= 6.00
SD= 0.00
min, max= [6.00, 6.00]

Spot tests
All negative

Sample B: contd.

Organic acid analysis

	n	points
Mevalonic acid / mevalonate lactone elevated	21	2

Mevalonolactone
n=6
median= 11128.00
mean= 18359.35
SD= 20086.43
min, max= [0.12, 55403.00]

Mevalonic acid
n=6
median= 3345.00
mean= 3212.52
SD= 2801.94
min, max= [15.16, 7971.00]

Interpretation

	n	Points
Mevalonate kinase deficiency	21	2

Sample C: Adenine phosphoribosyltransferase deficiency (APRTD); OMIM #614723

Patient details provided:
A 30 year old female with kidney and urinary stones. Urine collected at 33 years off treatment.

Further information
High levels of dihydroxy adenine found repeatedly which after starting allopurinol treatment fell dramatically (almost 10 fold). Over the last year she had to stop the treatment due to pregnancy when dihydroxy adenine was again very high. Urine collected at 33 years off treatment

Analytical performance*:*

The diagnosis relies on performing purine/pyrimidine analysis which was done by 15 labs. The key abnormality is the finding of increased 2,8, dihydroxy adenine and/or adenine, scored with 2 points (9/21 labs). One lab identified crystals suggestive of 2,8, dihydroxy adenine, scored with two points.

Interpretative proficiency:
The correct diagnosis is Adenine phosphoribosyltransferase deficiency (two points), correctly identified by 10 labs.
Recommendation to perform the correct analysis or to exclude APRT deficiency was scored with one point (6 labs).

Recommendations:
Following were given:
Repeat P/P analysis (n=4); Perform a test (P & P/ APRT / stone analysis) if not done (n=13); enzyme assay (n=6); mutation analysis (n=9); allopurinol treatment (n=4).

Overall impression
Overall proficiency of about 50% points to need to carry out or improve purine and pyrimidine analysis, especially since a few labs performed P/P analysis without detecting the key metabolite.

This was considered to be an educational sample, scores not to be taken into account in evaluating overall performance and therefore not to be eligible for critical error.

Analytical Details

Creatinine
n=20
median= 4.43
mean= 4.38
SD= 0.08
min, max= [3.50, 4.71]

pH
n=10
median= 6.00
mean= 6.15
SD= 0.11
min, max= [6.00, 7.00]

Spot tests

Nitrites: + (n=4), ++ (n=2), +++ (n=1).

Sample C: contd.

Purine / pyrimidine analysis (15 labs)

	n	points
Increased 2,8, dihydroxy adenine and/or adenine	9	2
Crystals suggesting 2,8, dihydroxy adenine	1	2
No abnormality	5	0

2,8-dihydroxyadenine
n=2
median= 118.90
mean= 118.90
SD= 98.90
min, max= [20.00, 217.80]

Adenine
n=6
median= 6.40
mean= 6.22
SD= 1.24
min, max= [4.00, 8.00]]

Uric acid was reported as low by 2, normal by 3 labs and elevated by one.

Interpretation

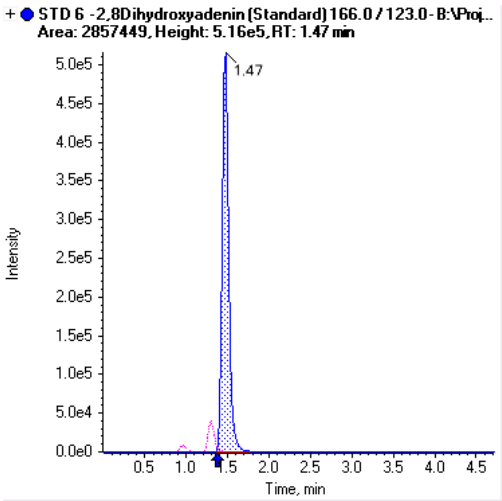
	n	Points
Adenine phosphoribosyltransferase deficiency	10	2
Recommendation to perform the correct analysis or to exclude APRT deficiency	6	1
Other IEM	1	0
No evidence of metabolic disorder	3	0
No diagnosis given	1	0

Sample C: contd.

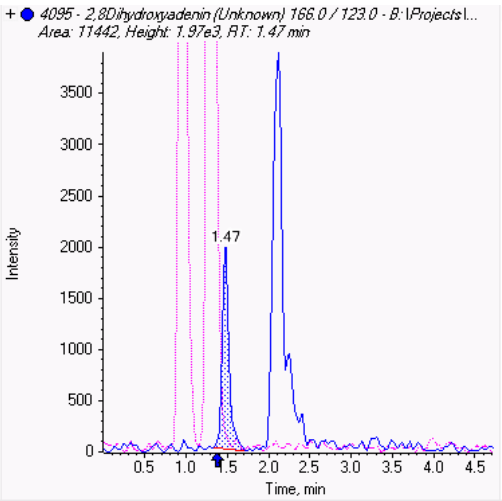
Purine / Pyrimidine Analysis – Tandem MS

2,8 – Dihydroxyadenine

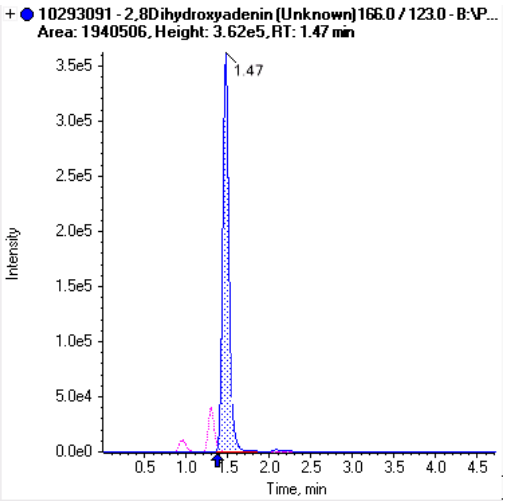
STD 50 µM



Healthy Control

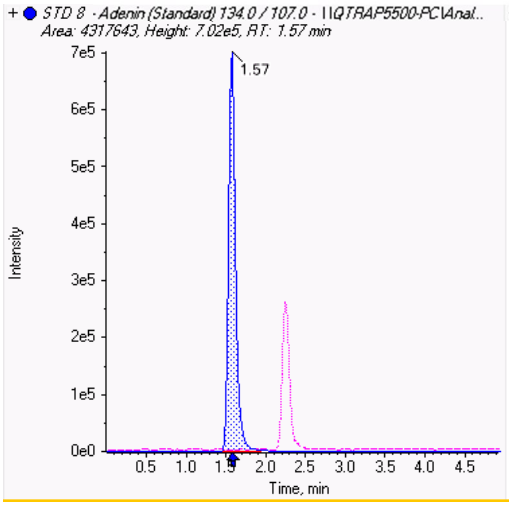


DPT 2017 C

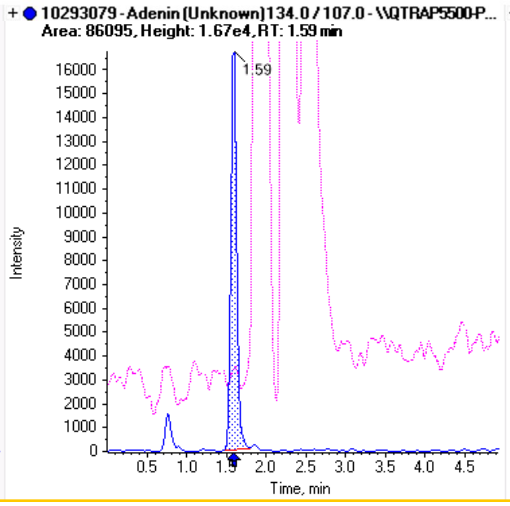


Adenine

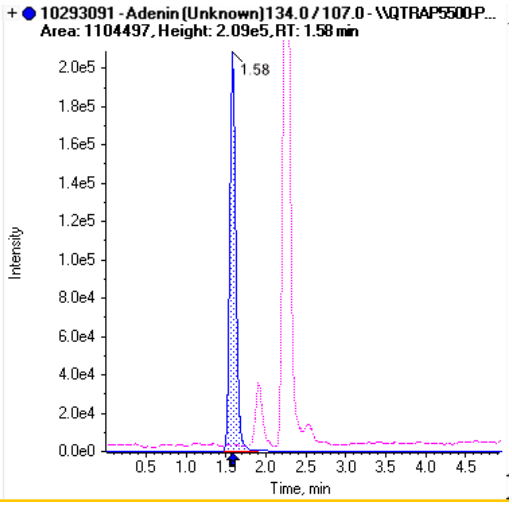
STD 18 µM



Healthy Control



DPT 2017 C



No detection of oxopurinol or allupurinol.

Sample D: No inborn error / on parenteral nutrition

Patient details provided:
Two month old male child with heart failure following operation. On non-specific treatment.

Further information
This male infant had been in the intensive care unit following a corrective heart operation due to heart failure (hypoplastic left ventricle). There was no suspicion of, or evidence for an inherited metabolic disorder. However management had included parenteral nutrition leading to a very prominent increased excretion of N-acetyl tyrosine which should be easy to identify in organic acid analysis by GC-MS.

Analytical performance:

Scoring of this sample was not straightforward and is therefore only provisional. Organic acid analysis was performed by all labs, 16 reported elevated N-acetyltyrosine (2 points). Three labs reported a normal result, one "mild pyruvic aciduria" and one mentioned unknown peaks. In addition 17 labs reported elevated GAG excretion, 4 labs did not perform this analysis. The five labs that did not report N-acetyltyrosine received one point for increased GAG excretion. Based on this scoring strategy overall analytical proficiency was 88%.

Interpretative proficiency:
The correct interpretation was considered to be no inborn error with elevated N-acetyltyrosine of exogenous origin, scored with two points (9 labs).
Analysis of GAGs by electrophoresis revealed no specific pattern suggestive of a particular MPS disorder. Thus follow up of this finding without indicating a specific MPS disorder received one point (8 labs). Over interpretation by diagnosis of a particular MPS disorder(s) received no point (1 lab) as did diagnosis of any other disorder (3 labs). Overall interpretative proficiency was 62%

Overall impression:

The sample was somewhat complex due to the non-specific elevation of GAG excretion. Nevertheless many labs correctly concluded no IEM but elevated N-acetyltyrosine of no consequence. Some labs over interpreted the GAG analysis which was possibly related to heparin treated as mentioned by a few labs. Finally the reliability of GAG quantitation in such a dilute sample must be questioned.

Recommendations:
The following were considered to be appropriate:
Questioning nutritional history and clinical features;
If correct diagnosis no follow up needed;
Repeat GAG analysis / or fractionation of GAGs, if not already performed;
Detailed enzyme /specific or general gene analysis should not be recommended.

This sample was considered by the SAB not to be eligible for critical error.

Analytical Details

Creatinine
n=21
median= 0.60
mean= 0.59
SD= 0
min, max= [0.49, 0.65]

pH
n=11
median= 6.00
mean= 5.59
SD= 0.24
min, max= [5.00, 6.00]

Sample D: contd.

Spot tests

All negative except positive “sulphides” reported by one lab.

Organic Acid Analysis (n= 21)

	n	points
N-acetyltyrosine	16	2
MPS increased (without N-acetyltyrosine)	5	1
Normal /pyruvate / unknown peaks	5	0

N-acetyltyrosine
n=3
median= 1132.00
mean= 1244.33
SD= 868.71
min, max= [241.00, 2360.00]

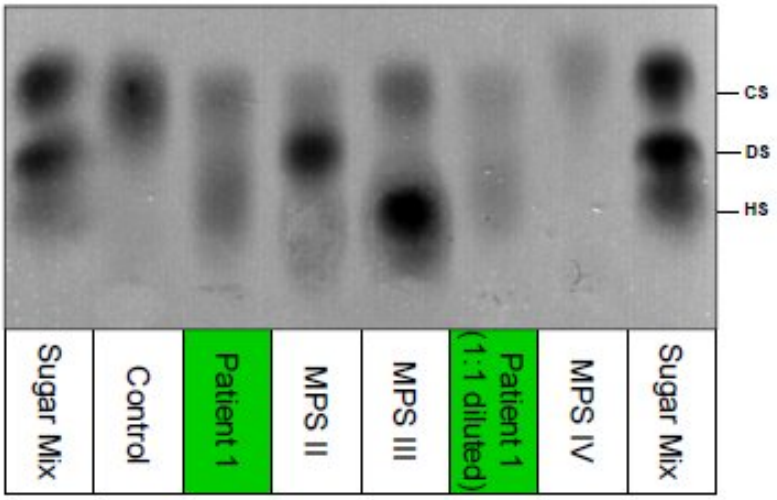
Interpretation

	n	Points
No inborn error, elevated N-acetyltyrosine of exogenous origin	9	2
MPS disorder not specified	8	1
Specified MPS disorder(s)	1	0
any other disorder	3	0

GAG electrophoresis

Date of Analysis: 04.07.2017 Operator: MT

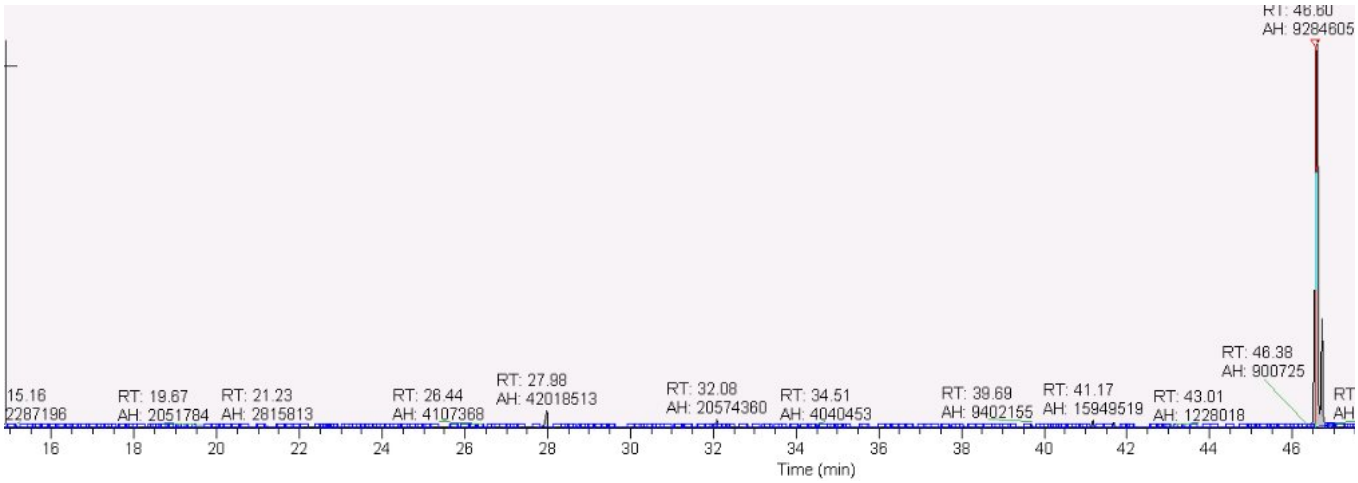
Patient 1: DPT 2017 (Brian Fowler), 6 Mths old Creat. 0.6 mmol/L

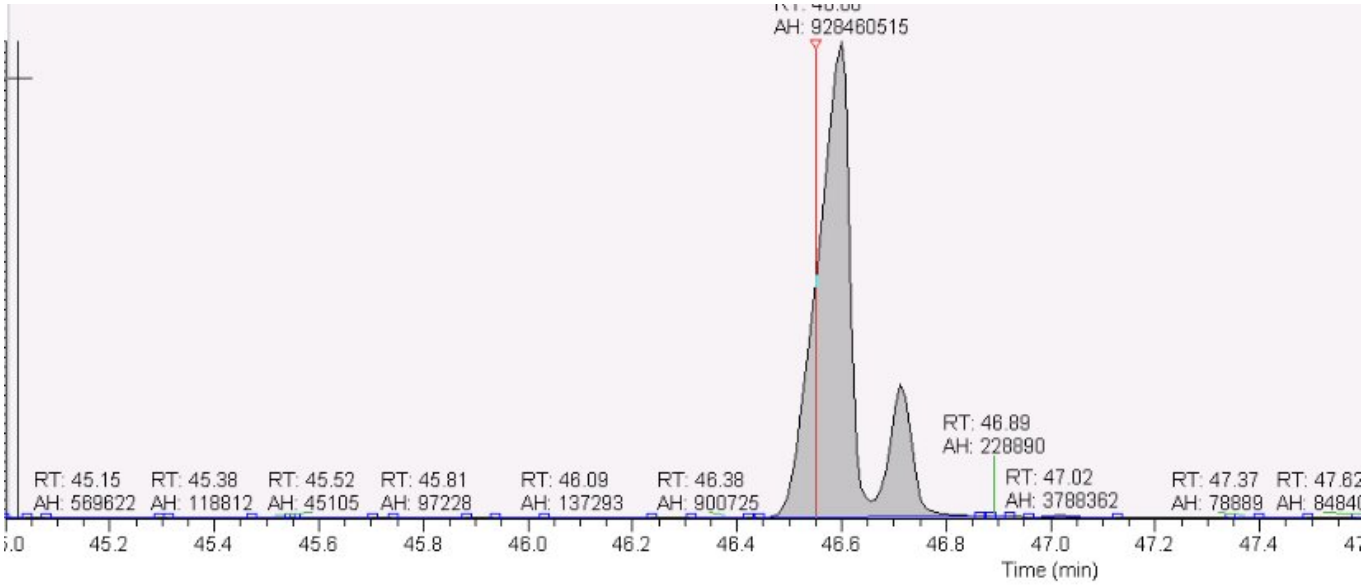


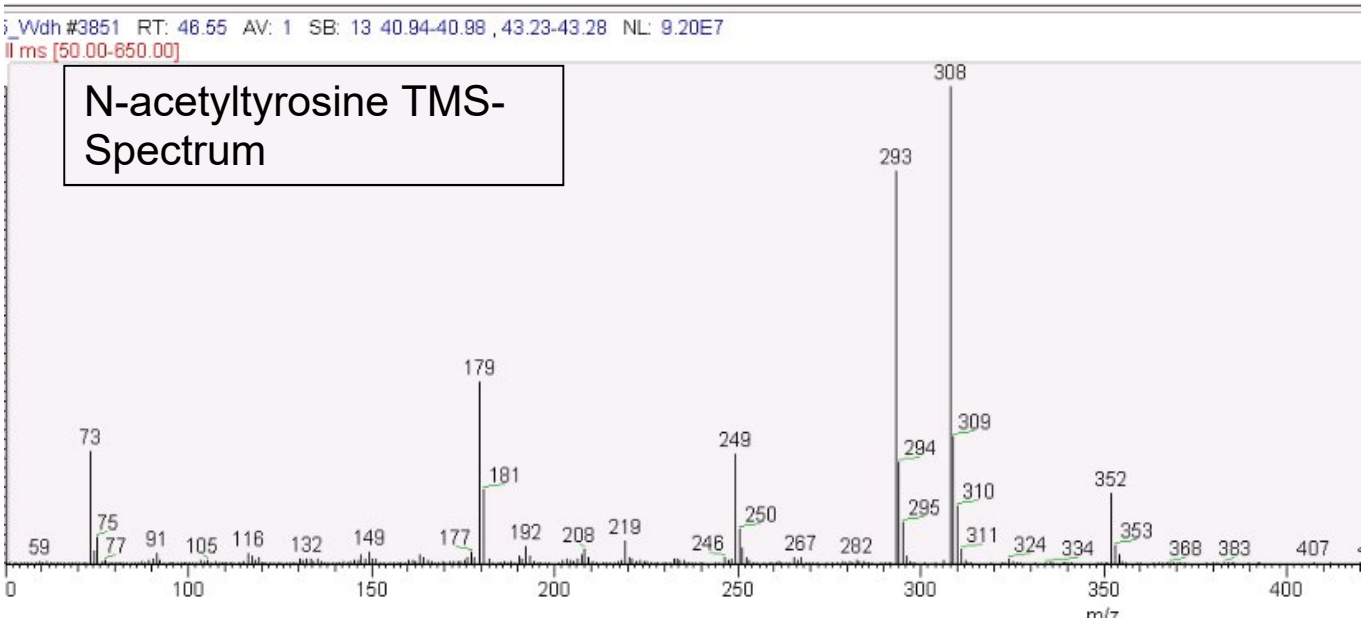
DMB-
115 mg/mmol Crea
Control upper limit 60

Sample D: contd.

Organic Acids-GCMS







Sample E: Prolidase deficiency,OMIM #170100.

Patient details provided:
Male child with developmental delay and skin rash at 2 years of age. Sample collected at 5 years no treatment
Further information
Initially we had not been allowed to see clinical details of the patient but the first sample was received at the age of two years for CDG analysis. The details quoted for this sample are certainly reminiscent of the typical symptoms of prolidase deficiency, namely skin lesions (ulcerations), mild intellectual disability and frequent infections.
Subsequently we received the following details about the child. The child had frequent infections, hepatosplenomegaly with elevated transaminases, pancytopenia and vasculitis from the age of one and half years. An inflammatory syndrome such as hemophagocytic lymphohistiocytosis was suspected. Prolidase deficiency was confirmed by molecular genetic analysis.

Analytical performance*:*
Dipeptides / iminopeptides pointing to the conclusion of prolidase deficiency was reported by 11 labs (2 points). Unknown or interfering peaks was reported by 4 labs, scored with one point. Increase of some individual amino acids (3 labs) or a normal amino acid pattern (3 labs) was scored with no points.

Interpretative performance:
Prolidase deficiency was correctly diagnosed by 11 labs (2 points). Further investigation by follow up with repeat amino acid analysis was considered by 7 labs (1 point).

Recommendations:
Confirmation of urine amino acid abnormality (4), analysis of the PEPD gene (9), prolidase assay (4) were correctly recommended. Biotinidase assay (2 labs) was not considered to be correct.

Overall impression:
Overall proficiency was moderate at 65%

The SAB considered that this sample was not eligible for critical error due to the low overall proficiency.

Analytical Details

Creatinine
n=21
median= 3.98
mean= 3.92
SD= 0.05
min, max= [3.33, 4.27]]

pH
n=11
median= 7.00
mean= 6.77
SD= 0.11
min, max= [6.00, 7.00]

Spot tests:
Glucose, + (n=4), ++ (n=1)
Protein, trace (n=2)
Reducing subs, trace (n=1)

Sample E: contd.

Amino acid analysis (n= 21)

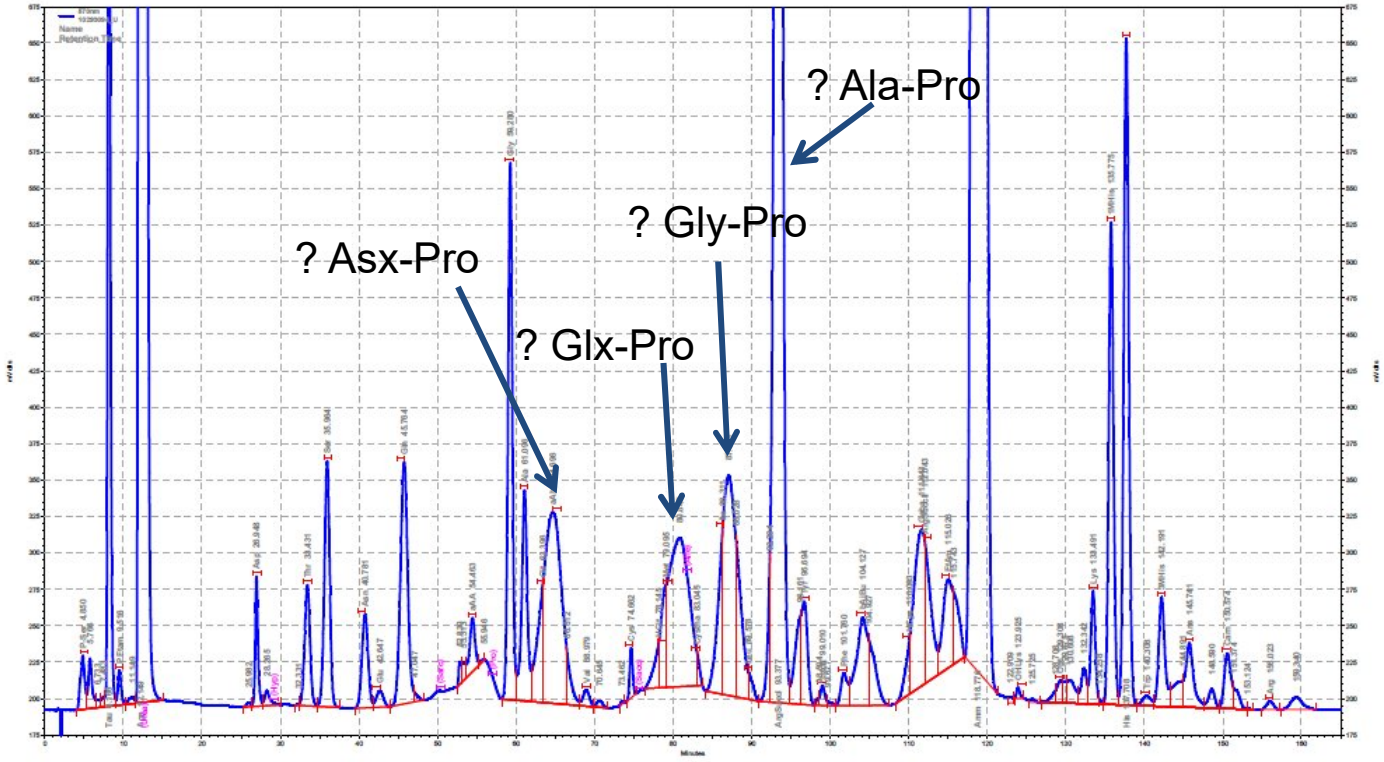
	n	points
Dipeptides / iminopeptides pointing to prolidase deficiency	11	2
Unknown or interfering peaks	4	1
increased individual amino acids	3	0
normal amino acid pattern	3	0

Glycylproline
n=2
median= 598.00
mean= 598.00
SD= 326.00
min, max= [272.00, 924.00]

Interpretation

	n	Points
Prolidase deficiency	11	2
Amino acid analysis follow up	7	1
No diagnosis/recommendation	3	0

Amino Acids – Ion-Exchange



For full description of retention times of proline dipeptides see: Ferreira CR and Cusman-Ozog K, JIMD Reports, DOI 10.1007/8904_2016_552

Sample F: Arginase deficiency, OMIM # 207800

Patient details provided:
Male child, 3 years of age with seizures, developmental delay and spastic paresis of lower extremities with failure to thrive. Sample collected at 10 years of age on treatment
.
Further information
A male child, 3 years of age with seizures, developmental delay and spastic paresis of lower extremities with failure to thrive. Sample collected at 10 years of age on protein restriction. Plasma arginine was 800-900 umol/L, NH3 100-120 umol/L.
A homozygous mutation in the arginase gene was confirmed (c.383A>G).

Analytical performance*:*

The sample proved to be somewhat complex due to the rather undramatic elevation of arginine together with a clear increase of homocitrulline and orotic acid. Following input from the participants' DPT meeting and input from the second assessor elevated arginine received 1 point (17 labs) and increase of either homocitrulline (18 labs) or orotic acid (21 labs) was scored with 1 point.

Interpretative proficiency:
Arginase deficiency as first or second diagnosis (7 labs) scored 2 points. A diagnosis of triple H syndrome or other urea cycle defect with a recommendation to measure plasma amino acids (11 labs) was also scored with 2 points but triple H syndrome without amino acid analysis recommendation received one point (3 labs). This is because the diagnostic pattern in the two disorders is difficult to distinguish in urine. There is great variability of levels of metabolites and increased homocitrulline has been reported in arginase deficiency (see below). Only a few labs commented on the clinical features that are typical for arginase deficiency.

Recommendations:
The most important recommendation is to measure plasma amino acids (15 labs) and ammonia (8), certainly before proceeding to molecular genetic analysis but the latter was suggested by 4 labs.

Overall impression:
Overall proficiency was high at 93%.

This sample was considered by the SAB not to be eligible for critical error.

Analytical Details

Creatinine
n=21
median= 5.88
mean= 5.78
SD= 0.34
min, max= [3.50, 6.34]

pH
n=11
median= 7.00
mean= 7.13
SD= 0.10
min, max= [7.00, 8.00]

Spot tests
Nitrites, tract (n=1)
Protein, trace (n=2)

Sample F: contd.

Amino acid analysis (n= 21)

	n	points
Arginine	17	2
Homocitrulline	17	1*
No abnormality	0	0

*1 point for homocitrulline or orotic acid

Arginine
n=16
median= 83.50
mean= 87.57
SD= 23.77
min, max= [53.00, 168.00]

Homocitrulline
n=11
median= 145.00
mean= 214.08
SD= 222.94
min, max= [92.00, 910.00]

Ornithine
n=13
median= 11.40
mean= 17.86
SD= 21.50
min, max= [8.00, 92.00]

Lysine
n=11
median= 160.28
mean= 177.28
SD= 59.26
min, max= [71.00, 323.00]

Organic Acid, Purine/ Pyrimidine analysis

	n	points
Orotic acid	21	1*

*1 point for homocitrulline or orotic acid

Orotic acid (Organic acids)
n=8
median= 351.15
mean= 349.28
SD= 211.02
min, max= [31.00, 803.00]

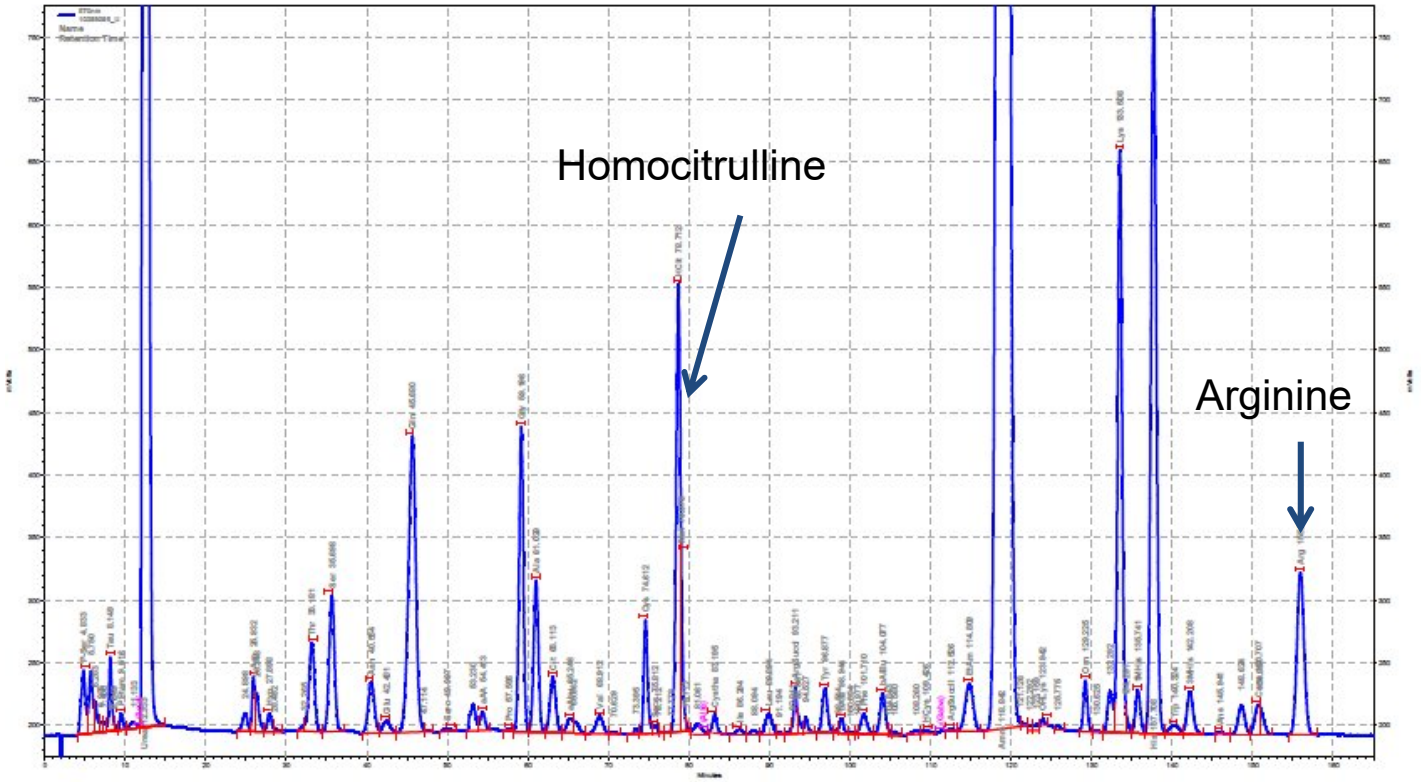
Orotic acid (Purine/Pyrimidines)
n=4
median= 394.90
mean= 394.20
SD= 80.49
min, max= [284.00, 503.00]

Interpretation

	n	Points
Arginase deficiency, first or second	7	2
Triple H syndrome /other UCD **with** amino acids recommended	11	2
Triple H syndrome /other UCD **without** amino acids recommended	3	1

Sample F: contd.

Amino Acids – Ion-Exchange



Homocitrulline in Hyperargininaemia

Increased homocitrulline has been reported in arginase deficiency by Kato et al. (J. Inher. Metab. Dis. 11 (1988) 261-265). Homocitrulline is thought to be formed by carbamylation of lysine by ornithine transcabamylase when carbamoyl phosphate accumulates which must be the case in this condition, supported by the finding of increased orotic acid.

8. Scores

Overall proficiency

Sample	Diagnosis	A (%)	I (%)	total (%)
A	Citrullinaemia	98	98	98
B	Mevalonic aciduria	100	100	100
C	APRT deficiency	48	62	55
D	No IEM	88	62	75
E	Prolidase deficiency	62	69	65
F	Arginase deficiency	90	93	92

Total scores

	Survey 1			Survey 2				
Lab No	A	B	C	D	E	F	Total All scored	C not scored
1	4	4	4	4	4	4	24	20
2	4	4	4	3	1	4	20	16
3	4	4	1	3	4	4	20	19
4	4	4	4	2	4	3	21	17
5	4	4	4	2	4	2	20	16
6	4	4	0	4	0	4	16	16
7	4	4	1	3	4	4	20	19
8	3	4	0	4	1	4	16	16
9	4	4	0	2	2	4	16	16
10	4	4	0	4	4	4	20	20
11	4	4	1	2	1	4	16	15
12	4	4	4	4	4	4	24	20
13	4	4	4	2	4	3	21	17
14	4	4	4	4	4	4	24	20
15	4	4	1	2	1	4	16	15
16	4	4	4	4	2	4	22	18
17	4	4	4	1	2	4	19	15
18	4	4	1	3	4	4	20	19
19	4	4	0	4	0	2	14	14
20	4	4	1	4	1	3	17	16
21	3	4	4	2	4	4	21	17

The scores proposed by us were evaluated by a second advisor and confirmed at the Scientific Advisory Board meeting in November 2016. At this meeting the cut off point for satisfactory performance was set (see below). Labs failing to reach this mark or making a critical error will receive a performance advice letter.

Detailed Scores: A,B,C

Lab no	Sample A Citrullinaemia			Sample B Mevalonic aciduria			Sample C APRT deficiency			
	A	I	Total	A	I	Total	A	I	Total	Total
1	2	2	4	2	2	4	2	2	4	12
2	2	2	4	2	2	4	2	2	4	12
3	2	2	4	2	2	4	0	1	1	9
4	2	2	4	2	2	4	2	2	4	12
5	2	2	4	2	2	4	2	2	4	12
6	2	2	4	2	2	4	0	0	0	8
7	2	2	4	2	2	4	0	1	1	9
8	1	2	3	2	2	4	0	0	0	7
9	2	2	4	2	2	4	0	0	0	8
10	2	2	4	2	2	4	0	0	0	8
11	2	2	4	2	2	4	0	1	1	9
12	2	2	4	2	2	4	2	2	4	12
13	2	2	4	2	2	4	2	2	4	12
14	2	2	4	2	2	4	2	2	4	12
15	2	2	4	2	2	4	0	1	1	9
16	2	2	4	2	2	4	2	2	4	12
17	2	2	4	2	2	4	2	2	4	12
18	2	2	4	2	2	4	0	1	1	9
19	2	2	4	2	2	4	0	0	0	8
20	2	2	4	2	2	4	0	1	1	9
21	2	1	3	2	2	4	2	2	4	11
ratio	41/42	41/42	82/84	42/42	42/42	**84/84**	20/42	26/42	**46/84**	
%	98%	98%	98%	100%	100%	100%	48%	62%	55%	

Detailed Scores: D,E,F

Lab no	Sample D No IEM			Sample E Prolidase def.			Sample F Arginase def.			
	A	I	Total	A	I	Total	A	I	Total	Total
1	2	2	4	2	2	4	2	2	4	12
2	2	1	3	0	1	1	2	2	4	8
3	2	1	3	2	2	4	2	2	4	11
4	1	1	2	2	2	4	1	2	3	9
5	2	0	2	2	2	4	1	1	2	8
6	2	2	4	0	0	0	2	2	4	8
7	2	1	3	2	2	4	2	2	4	11
8	2	2	4	0	1	1	2	2	4	9
9	1	1	2	1	1	2	2	2	4	8
10	2	2	4	2	2	4	2	2	4	12
11	2	0	2	0	1	1	2	2	4	7
12	2	2	4	2	2	4	2	2	4	12
13	2	0	2	2	2	4	2	1	3	9
14	2	2	4	2	2	4	2	2	4	12
15	1	1	2	0	1	1	2	2	4	7
16	2	2	4	1	1	2	2	2	4	10
17	1	0	1	1	1	2	2	2	4	7
18	2	1	3	2	2	4	2	2	4	11
19	2	2	4	0	0	0	1	1	2	6
20	2	2	4	1	0	1	1	2	3	8
21	1	1	2	2	2	4	2	2	4	10
ratio	37/42	26/42	**63/84**	26/42	29/42	**55/84**	38/42	39/42	**77/84**	
%	88%	62%	75%	62%	69%	65%	90%	93%	92%	

9. Assessment of performance

The Scientific Advisory Board of ERNDIM sets the level of good performance within a proficiency scheme. Letters of support to those laboratories with clear poor performance will be issued. The level for satisfactory performance for this year was set at the SAB meeting in November. A special meeting of scientific advisors took place in November 2012 to consider how to harmonise scoring within all our qualitative schemes and the question of introducing critical errors in our schemes. Here it was decided to incorporate recommendations into interpretation giving a 2 plus 2 scoring system. Also the concept of **critical error** was introduced in 2014**.** Thus labs failing to make a correct diagnosis of a sample considered as eligible for this category will be deemed not to have reached a satisfactory performance even if their total points for the year exceed the limit set at the SAB.
For 2017 samples **A and B** qualified as possible critical error.
Sample C was judged to be educational and its scores were not included in assessment of overall performance. Thus the level set for **satisfactory performance is twelve points** and below this is evaluated as unsatisfactory.
Note to educational samples: Samples may be classed as 'educational' in exceptional cases, e.g. when the metabolite pattern in a sample is particularly challenging and diagnosis is hard to reach or when non-standard methods are required. The Scientific Advisory Board decides whether a sample is classed as educational. When a sample, that has been classed as educational in an earlier survey, is circulated again it will be scored routinely and cannot be educational for a second time.

10. Annual meeting

The annual meeting of participants of this DPT centre took place, alongside those of the other four centres. during the special ERNDIM workshop in Manchester on November 21st. This was attended by 12 participants representing 8 centres. The agenda included:

- Discussion of organisational aspects, samples, delivery problems, and any requests for reporting improvements.
- Results of the individual samples, consensus on scoring and participants' performance.
- Overall performance and scores.
- Proposals for critical error samples.
- Future perspectives.
- DPT meeting in 2018: this is planned to be held at the SSIEM annual symposium in Athens, September 4th.

11. Changes planned for 2018

No changes are envisaged

12. Tentative schedule and fee in 2018

Sample distribution	February 5th, 2018
Start of analysis of Survey 2018/1	February 26th, 2018
Survey 2018/1 - Results submission	March 19th, 2018
Survey 2018/1 - Reports	May 15th, 2018
Start of analysis of Survey 2018/2	May 28th 2018
Survey 2018/2 – Results submission	June 18th, 2018
Survey 2018/2 - Reports	July 31st, 2018
Annual meeting of participants	SSIEM meeting Sept 2018
Annual Report 2018	December 2018

Fee was set at €437.

13. ERNDIM certificate of participation

A combined certificate of participation covering all EQA schemes will be provided to all participants who take part in any ERNDIM scheme. For the DPT scheme this certificate will indicate if results were submitted and whether satisfactory performance was achieved in the scheme.

Basel / Zürich, January 2018

Brian Fowler
Scientific advisor

Note: This annual report is intended for participants of the ERNDIM DPT-Switzerland scheme. The contents should not be used for any publication without agreement of the scheme advisor