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To

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ERNDIM QA Scheme for Qualitative Blood Spot Acylcarnitine Analysis

Annual Report 2014

Participation

The geographical distributions of the active participants of the quality assurance scheme organized and distributed through the centre of Heidelberg in 2014 are shown in Table 1. London and Heidelberg participate in each other´s scheme and the two centers work closely together under the auspices of the ERNDIM Scientific Advisory Committee.

Country	Number of laboratories
Argentina	3
Austria	1
Belgium	6
Brazil	1
Bulgaria	1
China	4
Czech Republic	2
France	18
Germany	7
Greece	1
Hong Kong S.A.R.	1
Lebanon	1
Luxembourg	1
Slovakia	1
Switzerland	3
The Netherlands	8
Turkey	1
United Kingdom	2
Total	62

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Samples and results

Two sets of three blood spot samples (total 6; sample number 23A, 23B, 23C, 24A, 24B, 24C) were distributed to 62 laboratories.

Nine participants did not answer to any of the two circulations. Seven laboratories returned results only for one circulation.

Table 2: Receipt of results

Circulation	In time returns	Late returns	Total
1. circulation	49	2	51
2. circulation	46	3	49

Shipment of the samples

Blood spot samples prepared on Whatman 903™ specimen collection paper were shipped on 19 September 2014 and on 17 November 2014.

Table 3: Distribution of scores for individual samples (laboratories making returns)

		4	3	2	1	0
Sample 23A	Isovaleric aciduria	51				
Sample 23B	Cobalamine A deficiency	49	1			1
Sample 23C	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	51				
Sample 24A	Propionic acidaemia	49				
Sample 24B	Glutaric aciduria type I (low excreter)	47			1	1
Sample 24C	Normal profile	46		3		

Comments on performance

The overall performance (analytical and diagnostic) for sample 23A (**isovaleric aciduria)** was at 100%. All participants clearly detected increased C5-carnitine.

For sample 23B minimal requirement for good analytical performance was detecting elevated amounts of C3-carnitine. This was achieved by 98% of the participants. Based on this analytical finding 37% of the participants diagnosed **methylmalonic acidaemia**, 35% diagnosed **propionic acidaemia**, 20% **disorders of cobalamin metabolism**. Three laboratories gave these diagnoses only as an alternative to a normal profile.
One participant found the acyl carnitine profile to be normal.

Analytical and diagnostic performance for **medium-chain acyl-CoA dehydrogenase (MCAD) deficiency** (sample 23C) was 100%. All laboratories clearly detected increased C8-carnitine.

The overall performance (analytical and diagnostic) for sample 24A (**propionic aciduria)** was also 100%. Elevated C3-carnitine was detected by all participants.

Sample 24B was collected from a low excreting patient with **glutaric aciduria type I.** The analytical performance in detecting glutarylcarnitine (C5DC) was 90%. Two laboratories did not find any abnormalities. So diagnostic performance was only 94% with one laboratory diagnosing carnitine palmitoyltransferase I (CPT1) deficiency, and another one giving glutaric aciduria type I as an alternative diagnosis.

The **normal control** *sample 24C* was correctly identified by 90% of the participants. Three laboratories diagnosed either MCAD deficiency, a possible heterozygote of carnitine transporter deficiency or a disorder in the metabolism of vitamin B_{12}.

Scoring scheme

In the process of ongoing accreditation of the ERNDIM organization there is a need for harmonization of performance assessment within the qualitative schemes (see ERNDIM 'Newsletter Spring 2013' at www.erndim.org).

In 2013 we changed the scoring system from the former scale (-2, -1, 0, +1, +2) to the four-point system (+1, +2, + 3, +4) which is used also in the DPT schemes. In this system a maximum of two points is given each for analytical results and interpretation, with the latter including suggestions for further testing/actions.

The total score achievable for a single circulation of three samples is twelve. The maximal achievable score, full points for the year is twenty-four for the whole sample set of six samples in the year.

To obtain satisfactory performance a score of 16 or more should be achieved on two returns. Laboratories that participate only in one circulation are treated as non-submitters.

Another criteria for satisfactory performance will be the absence of any "critical error" which is defined as an error resulting from seriously misleading analytical findings and /or interpretations with serious clinical consequences for the patient.

The participants´ cumulative scores are shown in table 4. Cumulative scores are the scores for the whole year.

This year forty-one participants got full marks. This is 89% of all participants that returned results for both circulations, and 66% of all registered participants.

Table 4: cumulative total scores 2014 (all registered laboratories that returned results for both circulations)

	Percent of participants	
Cumulative scores	**2014**	**2013** (maximal achievable score was 20)
24	89.2	Not defined
23	-	Not defined
22	4.3	Not defined
21	4.3	Not defined
20	2.2	71.7
19	-	6.5
18	-	6.5
17	-	8.7
16	-	6.5
15	-	-
14	-	-
13	-	-
12	-	-
11	-	-
10	-	-
9	-	-
8	-	-
7	-	-
6	-	-
5	-	-
4	-	-
3	-	-
2	-	-
1	-	-
0	-	-
Number of all participants	62	60
Number of Nonresponders	16	14

Your individual scores for Sample 23A – 24C:

Sample 23A

Sample 23B

Sample 23C

Sample 24A

Sample 24B

Sample 24C

Your total score 2014

Your total score for 2014 was:

Your number of returns in 2014 was:

General comments

We would like to point out here that we are not able to accept returns sent in after the report for the corresponding circulation has been mailed because this would not be compatible with the overall intention of the scheme. We are conscious of the fact that posted results could get lost on a variety of ways. Therefore it would be a good advice to send in results on more than one route (e.g. FAX and email, regular mail and FAX or email).

Appeal for contributing samples:

To keep the acylcarnitine scheme running we would like to encourage all participants to support us with samples. We need blood spots or whole blood. The shipping costs will be covered by us.

Please contact us under claus-dieter.langhans@med-uni-heidelberg.de for the details.

Yours sincerely,

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