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Kinderheilkunde I
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Universitätsklinik für Kinder- und Jugendmedizin

ERNDIM QA Scheme for qualitative blood spot acylcarnitine analysis

Annual Report 2010

Participation

The geographical distributions of the active participants of the quality assurance scheme organized and distributed through the centre of Heidelberg in 2010 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
Agentina	2
Austria	2
Belgium	5
Brazil	1
Czech Republic	3
France	2
Germany	11
Greece	6
Lebanon	1
Luxembourg	1
Switzerland	1
The Netherlands	8
Turkey	2
United Kingdom	2
Total	**50**

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Samples and results

Two sets of three blood spot samples (total 6; sample number 15A, 15B, 15C, 16A, 16B, 16C) were distributed to 50 laboratories.

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

Table 2: Receipt of results

Circulation	Number of returns	Late returns
1. circulation	41	1
2. circulation	43	1

Shipment of the samples

Blood spot samples prepared on Whatman 903™ specimen collection paper were shipped on 25th November 2010 and on 31st August 2010.

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

		-2	-1	0	1	2
Sample 15A	Glutaric aciduria type I	2			2	37
Sample 15B	Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)	10		4	3	24
Sample 15C	Methylmalonic aciduria			1		40
Sample 16A	Isovaleric aciduria					43
Sample 16B	Normal profile	8	1		1	33
Sample 16C	Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)			1		42

Comments on performance

The detection rate for the classical organoacidopathies **methylmalonic aciduria (#15C)** and **isovaleric aciduria (#16A)** was 98% and 100% respectively.

In case of **glutaric aciduria type I (#15A)** the analytical performance in detecting increased glutaryl carnitine (C5DC) was 93% and parallels the interpretative proficiency. False negative results are at 7% and could be attributed to failures in identifying increased C5DC.

In the group of fatty acid oxidation disorders the overall performance varied greatly between the disorders and was at 98% for **medium-chain acyl-CoA dehydrogenase (MCAD) deficiency (#16C)**.

In contrast for **long-chain hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency (#15B)** analytical and diagnostic interpretation was at 66% with 24% of false negative results. A possible reason for this might be that the indicative metabolites were only slightly increased in #15B due to blood sampling in a compensated situation.

The overall performance in identifying a **normal acylcarnitine profile (#16B)** was 77% whereas 23% reported an abnormal profile. The normal profile was combined with the clinical information suggesting e.g. CPT II deficiency and this was the most common suspicion.

Scoring scheme

Individual returns for each sample were scored on the scale

2	Correct/satisfactory
1	Helpful but incomplete
0	Unhelpful / failing to return a result
-1	Slightly misleading
-2	Misleading

The ERNDIM organisation provides a single "Certificate" to cover participation and performance in all its schemes.
For the "Qualitative Acylcarnitine Scheme" we adopted the criteria to define "Participation" and "Satisfactory Performance" from the well-established system of the "Qualitative Organic Acid Scheme".
"Participation" will be defined as requiring all two returns during a year and "Satisfactory Performance" as obtaining a score of 7 or more out of maximum score 12.
We are aware that these criteria are rather arbitrary but we are convinced that they will represent the different contexts in which the participants are working.

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

This year seventeen participants (34%) got full marks!

Five laboratories i.e. 12% of the active participants failed to fulfill the above mentioned criteria for "Satisfactory Performance".

Table 4: cumulative total scores 2010 (all registered laboratories)

	Number of laboratories
Cumulative scores	**2010**
12	17
11	2
10	4
9	0
8	11
7	4
6	1
5	0
4	1
3	1
2	1
1	0
0	8

Your individual scores for #Sample 15A – 16C:

Sample #15A
Sample #15B
Sample #15C

Sample #16A
Sample #16B
Sample #16C

Your total score 2010

Your total score for 2010 was:

Your number of returns in 2010 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).

Yours sincerely,

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