

**ERNDIM QA Scheme for
qualitative urinary organic acid
analysis**

Annual Report 2008

Participation

The geographical distributions of the active participants in 2008 are shown in Table 1. Sheffield and Heidelberg participate in each other's scheme and the two centres work closely together under the auspices of the ERNDIM Scientific Advisory Committee.

Table 1: Geographical distribution of participants

Country	Number of laboratories	Country	Number of laboratories
Austria	3	Luxemburg	1
Belgium	1	Norway	1
Canada	6	Philippines	1
Croatia	1	Poland	1
Cypres	1	Saudi Arabia	3
Czech Republic	2	Slovakia	1
Denmark	1	Slovenia	1
Estonia	1	Spain	2
France	2	Sweden	2
Germany	11	Switzerland	3
Greece	1	The Netherlands	10
Hungary	1	United Kingdom	1
Israel	1	USA	12
Italy	10	Vatican City	1
Latvia	1		

Im Neuenheimer Feld 153
69120 Heidelberg

Stoffwechsellabor:

Fon +49 (0)6221 56 8276

8423

Fax +49 (0)6221 56 5565

Stoffwechselklinik und -ambulanz:

Fon +49 (0)6221 56 2319 (Anmeldung)

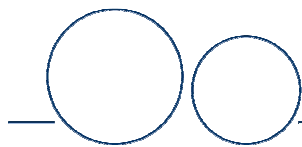
2311 (Information)

Neugeborenenenscreening:

Fon +49 (0)6221 56 8278

stoffwechsellabor@uni-hd.de

www.stoffwechsel.uni-hd.de



Samples and results

Three sets of three samples (total 9; sample number 160 --168) were distributed to 83 laboratories.

Three participants did not answer to any of the three circulations. Nine laboratories returned results for two circulations, three for only one.

Circulation	Number of returns	Late returns
1. circulation	77	3
2. circulation	75	4
3. circulation	74	2

Shipment of the samples

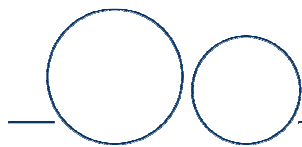
As the years before we sent out the samples for all three circulations together. This is only for organisation reasons and to keep the costs for participating in this scheme as low as possible.

Please remember, the idea of the scheme is to measure the samples evenly spread over the year and report the results near the closing date!

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

		-2	0	1	2
Sample 160	Normal pattern	1	-	1	75
Sample 161	Isovaleric aciduria	-	-	-	77
Sample 162	Mevalonic aciduria	-	-	-	77
Sample 163	Normal pattern	3	1	-	71
Sample 164	Normal pattern	2	1	4	68
Sample 165	3-hydroxy-3-methylglutaryl CoA lyase deficiency	-	1	1	73
Sample 166	Tyrosinaemia type I	7	6 ^{*)}	32 ^{*)}	29
Sample 167	Normal pattern	-	1	-	73
Sample 168	Maple syrup urine disease (MSUD)	4	-	1	69

*) updated scores of the individual report



Scoring scheme

Individual returns for each sample were scored on the scale

2	Correct/satisfactory
1	helpful but incomplete
0	unhelpful
-2	misleading

The ERNDIM organisation is moving towards providing a single “Certificate” to cover participation and performance in all its schemes. The scheme organizers of the “Qualitative Organic Acid Scheme” in Sheffield and Heidelberg agreed on criteria to define “Participation” and “Satisfactory Performance”.

We are aware that these criteria are rather arbitrary but we are convinced that they will represent all the different contexts in which the participants are working.

So “Participation” will be defined as requiring at least two returns during a year and “Satisfactory Performance” as obtaining a score of 11 or more based on three returns (out of maximum 18). When two returns have been received a score of 7 or more (in this case possible maximum score 12) would be considered satisfactory.

Comments on performance

In 2008 nearly all participants clearly diagnosed **isovaleric aciduria**, **mevalonic aciduria** and **3-hydroxy-3-methylglutaryl CoA lyase deficiency**.

With **maple syrup urine disease** a few laboratories seem to have difficulties in either detecting the diagnostically relevant metabolites or estimating significantly increases in these metabolite excretions.

Misinterpretation of analytical findings was not a major problem in all laboratories and was in this year’s circulations mainly associated with the identification of **normal urine profiles**. The overestimation of metabolites like a moderately prominent lactate peak in **#sample 164** or tricarboxylic acid cycle constituents in **#sample 163** may reflect different experiences of the participating laboratories.

In case of **tyrosinaemia type I** most of the participants identified metabolites typical of liver diseases. The key metabolite succinylacetone which was present in this urine sample only in trace amounts was detected by nearly forty percent of the laboratories.

The participants’ cumulative scores are shown in table 4. Cumulative scores are the scores for the whole year. This year twenty-one participants (29%) got full marks!

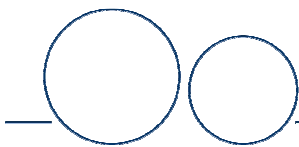


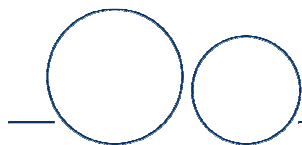
Table 4: cumulative total scores 2008 - 2005

Cumulative scores	Number of laboratories			
	2008	2007	2006	2005
18	21	55	16	25
17	26	4	10	4
16	5	-	10	12
15	1	-	4	1
14	11	2	12	6
13	2	-	2	2
12	6	7	5	6
11	3	2	-	1
10	1	1	4	3
9	-	-	1	1
8	1	1	-	-
7	1	-	-	1
6	1	3	3	-
5	-	-	-	-
4	-	-	-	-
3	-	-	-	-
2	-	-	-	2
1	-	-	-	-
0	4	3	4	3

Your total score 2008

Your total score for 2008 was:

Your number of returns in 2008 was:



General comments

We would just 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 in 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).

Special thank for the laboratories that supported us last year with samples. This is critical for the success of the program and will keep the scheme interesting. **It is most appreciated that you will continue to support us with urine from patients. Please send us at least 250 ml urine of any interesting patients you may have. We will cover the costs.**

Yours sincerely,

Dr. C. D. Langhans

*Laboratory of Metabolic
Diseases*

Dr. V. Peters

*Laboratory of Metabolic
Diseases*

Prof. Dr. G. F. Hoffmann

*Director
Department of General
Paediatrics*