

UniversitätsKlinikum Heidelberg

Universitätsklinik für Kinder- und Jugendmedizin Stoffwechselzentrum Heidelberg Stoffwechsellabor Im Neuenheimer Feld 153 | 69120 Heidelberg Stoffwechselzentrum Heidelberg Stoffwechsellabor

Kinderheilkunde I (Schwerpunkt: Allgemeine Pädiatrie, Stoffwechsel, Gastroenterologie u. Nephrologie) Prof. Dr. med. G.F. Hoffmann Ärztl. Direktor

Universitätsklinik für Kinder- und Jugendmedizin

ERNDIM QA Scheme for qualitative urinary organic acid analysis

Annual Report 2009

Participation

The geographical distributions of the active participants of the quality assurance scheme organized and distributed through the centre of Heidelberg in 2009 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	2	Norway	1		
Belgium	1	Philippines	1		
Canada	6	Poland	1		
Croatia	1	Saudi Arabia	2		
Cypres	1	Slovakia	1		
Czech Republic	2	Slovenia	1		
Denmark	1	Spain	3		
Estonia	1	Sweden	2		
France	2	Switzerland	3		
Germany	12	The Netherlands	10		
Greece	1	United Kingdom	1		
Hungary	1	USA	14		
Italy	11	Vatican City	1		
Latvia	1				
Luxembourg	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) Neugeborenenscreening: 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 169 --177) were distributed to 85 laboratories.

Six participants did not answer to any of the three circulations. Ten laboratories returned results for two circulations, one for only one circulation.

Table 2: Receipt of results					
Circulation	Number of returns	Late returns			
1. circulation	77	2			
2. circulation	74	3			
3. circulation	74	4			

Shipment of the samples

As the years before we sent out the samples for all three circulations together. This is only for organisation reasons especially 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 169	Alkaptonuria	2	-		76
Sample 170	Normal pattern	-	-	-	78
Sample 171	Aminoacylase I deficiency	39	3	3	33
Sample 172	Methylmalonic aciduria	-	-	-	74
Sample 173	Normal pattern	-	-	-	74
Sample 174	Canavan disease	1	-	1	72
Sample 175	Normal pattern	-	-	1	73
Sample 176	Phenylketonuria	1	-	-	73
Sample 177	Normal pattern	3	1	1	69

Scoring scheme

Individual returns for each sample were scored on the scale

- 2 Correct/satisfactory
- 1 helpful but incomplete
- o 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 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) is satisfactory.

Comments on performance

The overall performance in diagnosing **alkaptonuria**, **Canavan disease**, **methylmalonic aciduria** and **phenylketonuria** was excellent for the majority of laboratories. **Normal organic acid profile**s were also clearly identified by nearly all participants.

The most challenging sample in 2009 was **#sample 171**. This urine originated from a patient with **aminoacylase I deficieny**. Analytical performance in detecting the N-acetylated amino acid varied greatly between the laboratories. Only fifty-eight percent of the active participants clearly identified these compounds possibly due to different experiences of the participating laboratories.

The participants' cumulative scores are shown in table 4. Cumulative scores are the scores for the whole year. This year twenty-seven participants (32%) got full marks!



	Number of laboratories				
Cumulative scores	2009	2008	2007	2006	2005
18	27	21	55	16	25
17	3	26	4	10	4
16	2	5	-	10	12
15		1	-	4	1
14	31	11	2	12	6
13	1	2	-	2	2
12	6	6	7	5	6
11		3	2	-	1
10	1	1	1	4	3
9	1	-	-	1	1
8	4	1	1	-	-
7		1	-	-	1
6	2	1	3	3	-
5		-	-	-	-
4	1	-	-	-	-
3		-	-	-	-
2		-	-	-	2
1		-	-	-	-
0	6	4	3	4	3

Table 4: cumulative total scores 2009 - 2005

Your total score 2009

Your total score for 2009 was: Your number of returns in 2009 was:

> stoffwechsellabor@uni-hd.de www.stoffwechsel.uni-hd.de

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 on a variety of ways. Therefore it would be a good advice to send in results on more than one routes (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.

In 2009 samples for the QOA scheme were kindly contributed by

Prof. Dr. J. O. Sass, Freiburg, Germany

Yours sincerely,

tanglu

Potes

by un-

Dr. C. D. Langhans

Dr. V. Peters

Laboratory of Metabolic Diseases

Laboratory of Metabolic Diseases Prof. Dr. G. F. Hoffmann

Director Department of General Paediatrics

> stoffwechsellabor@uni-hd.de www.stoffwechsel.uni-hd.de